A distinctive childhood epilepsy syndrome

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DESCRIPTION

A 9-year-old boy presented with recurrent stereotypic paroxysmal movements of upper limbs and head shaking for past 2 years. Each episode had an abrupt onset and termination and was characterised by a rhythmic jerking of arms, shoulders and the head. The event was associated with vacant stare and lasted for 5–30 s. These events were not associated with eyelid blinking, perioral twitching or loss of postural tone. He had complete amnesia for these events. The frequency of these events progressively increased to 15–20 episodes per day. There was no recent change in behaviour, decline in school performance or any other affected family member. On examination, his anthropometry, neurological and systemic examination were unremarkable. During the clinical examination, the typical event was precipitated by hyperventilation (video 1). Ictal electroencephalogram showed generalised,

Figure 1 Awake ictal electroencephalogram (common average montage, sensitivity: 30 μ V/mm, sweep speed: 30 mm/s) showing abrupt onset of monomorphic bifrontal-dominant generalised 3- to 3.5-Hz spike-and-wave discharges with dart-and-dome that coincides with myoclonic jerks and has an abrupt offset.

Video 1 The typical event was precipitated by hyperventilation and characterised by abrupt cessation of activity followed by jerks of upper limbs and head. Child had complete amnesia of the event.

synchronous 3 Hz spike and slow-wave discharge with abrupt onset and offset (figure 1). An electroclinical diagnosis of epilepsy with myoclonic absences (EMA) was considered. He attained good seizure control with sodium valproate (25 mg/kg/day) and lamotrigine (3 mg/kg/day), and was seizure free till his last follow-up (16 months).

The syndrome of EMA is also called as the Tassinari syndrome. It was first described by Tassinari and colleagues¹ in 1969 and characterised by jerks of limbs, head or both with loss of awareness. The peak age of the onset of seizures is 3–7 years. Affected children have normal intellect before the onset of seizures, but most of the children have cognitive slowing or school difficulties after the onset of symptoms.² Few children may also have generalised tonic-clonic seizures. The frequency of myoclonic absences varies from 1 to 100 episodes per day and they can be precipitated by hyperventilation. Ictal EEG typically shows generalised spike-wave discharges of 3–3.5 Hz frequencies.³ Distinct

Learning points

- ▶ Tassinari syndrome also called as epilepsy with myoclonic absences and is characterised by jerks of limbs, head or both with loss of awareness.
- ▶ Affected children have normal intellect before the onset of seizures, but most of them have cognitive slowing or school difficulties after the onset of symptoms.
- ▶ The long-term prognosis of Tassinari syndrome is guarded and most of the individuals require polytherapy.

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from other genetic generalised epilepsies, the long-term prognosis is guarded and most of the individuals require polytherapy with valproic acid, ethosuximide, lamotrigine, levetiracetam and benzodiazepines.²

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