Brain & Development 36 (2014) 892–898

BRAIN & DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

www.elsevier.com/locate/braindev

Original article

Epilepsy with myoclonic absences in siblings

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Received 11 June 2013; received in revised form 16 December 2013; accepted 17 December 2013

Abstract

Background: Epilepsy with myoclonic absences (EMAs) is a distinct form of childhood epilepsy characterized by a peculiar seizure type that identifies this condition. *Purpose*: To describe the clinical, electroencephalographic features, treatment strategies and outcome in this first case series of two siblings with normal intelligence presenting with EMAs. *Materials and methods*: Both siblings underwent video-polygraphic investigations (simultaneous recording of electroencephalogram [EEG] and electromyogram [EMG] from deltoids), high-resolution magnetic resonance imaging (MRI), karyotyping, neuropsychological evaluation and language assessment. *Results*: Both the children had a mean age of onset of prototype seizures by 3.5 years. Myoclonic absences (MAs) were characterized by rhythmic, bilateral, synchronous, symmetric 3-Hz spike–wave discharges, associated with EMG myoclonic bursts at 3 Hz, superimposed on a progressively increasing tonic muscle contraction. The interictal EEG showed a normal background activity with bursts of generalized spike and waves (SWs) as well as rare focal SWs independently over bilateral temporal and frontal regions. Increase in the seizure frequency from 5 to 100/day was observed due to use of carbamazepine and phenobarbitone which decreased with its withdrawal and introduction of valproate. Though lamotrigine was given as an add on to valproate, it did not benefit them and was therefore replaced by topiramate at 3.5 mg/kg/day which has maintained them on remission at one year follow up. *Conclusions*: Recognition of this ictal pattern allows identification and differentiation of EMAs from other seizure types. Idiopathic and symptomatic EMAs need to be differentiated from childhood absence epilepsy with myoclonia. MAs are worsened by drugs like carbamazepine while valproate either alone or in combination with topiramate (preferred to lamotrigine) gives excellent outcome.

Keywords: Tassinari; EMAs; Valproate; Ethosuximide; 3 Hz; Polygraphic VEEG; Topiramate

1. Introduction

Epilepsy with myoclonic absences (EMAs) is characterized by absence seizures associated with rhythmic, bilateral proximal myoclonic jerks of upper extremities. The diagnosis is based on clinical observation and ictal video electroencephalograph (EEG) recordings. Demonstration of myoclonic absences (MAs) is essential for the diagnosis. In the proposed diagnostic scheme by the International League Against Epilepsy (ILAE) [1], it has been tentatively placed among the idiopathic generalized epilepsies.

2. Materials and methods

2.1. Subjects

Both the subjects who are siblings presented with seizures to our institute, which is a tertiary referral centre

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http://dx.doi.org/10.1016/j.braindev.2013.12.004

for neurological diseases in South India. They underwent video-polygraphic investigations, high-resolution magnetic resonance imaging (MRI), karyotyping, neuropsychological evaluation and language assessment.

2.2. Electroencephalography (EEG)

All recordings were carried out on a 16-channel digital EEG acquisition system (NicVue, Nicolet-Viking, USA); with the scalp electrodes placed according to the International 10–20 system. Both patients with EMAs also underwent a video-polygraphic investigation (simultaneous recording of electroencephalogram [EEG] and electromyogram [EMG] from deltoids). The distribution of interictal epileptiform discharges (IEDs) during prolonged video-EEG monitoring was assessed by visual analysis of the entire acquired data.

2.3. Neuroimaging

Both the patients underwent a 1.5 Tesla magnetic resonance imaging (MRI) (1.5 T MRI, Avanto, TIM SQ engine, Siemens, Erlangen, Germany).

2.4. Language and neuropsychological assessment

Malin's Intelligence Scale for Indian children, which is a validated Indian adaptation of Wechsler's Intelligence Scale for children, was used to assess the intelligence quotient (IQ) for children [2]. Language was assessed by the local adaptation of the Receptive Expressive Emergent Language Scale (REELS) and its extended version [3].

2.5. Follow-up and outcome assessment

Both the siblings were followed up at three-monthly intervals. The seizure and cognitive outcomes were assessed at each follow-up visit.

3. Results

3.1. Clinical, seizure and imaging characteristics

The siblings were born of nonconsanguinous parentage and had normal growth and developmental milestones. The median age at seizure onset was 3.5 years. Myoclonic absence seizures were characterized by recurrent episodes of rhythmic myoclonic jerks mainly at proximal shoulder associated with progressive elevation of the arms with unresponsiveness lasting for 10 s (Video 1).

The onset and offset were abrupt with no postictal confusion. They had no eyelid or perioral myoclonia. The seizures were predominantly diurnal especially in the early morning hours precipitated by hyperventilation, lasting for 10–15 s at a frequency of 4–5 per day. The girl had a past history of a single generalized tonic clonic seizure at the age of 3 years which was not associated with fever. Other than her younger brother who had EMAs no one else in the family was affected (Table 1). MRI including high resolution three-dimensional fast fluid attenuated inversion recovery (3D FLAIR) images with thin cuts were normal in both the cases.

3.2. EEG characteristics

The interictal EEG showed a normal background activity with bursts of generalized spike and waves (SWs) as well as focal SWs independently over bilateral temporal regions in the sister while her brother showed independent IEDs over bilateral frontal regions (Figs. 1–3). The ictal EEG consisted of rhythmic SW discharges at 3 Hz, which are bilateral, synchronous, and symmetric, as observed in typical absence seizures. The onset and the end of SWs were abrupt. Polygraphic (EEG–EMG) recording disclosed the appearance of bilateral myoclonias, at the same frequency as the SWs, which began around 1000 ms after the onset of EEG paroxys-

Table 1
Electro clinical profile, treatment and outcome of siblings with EMAs.

Current age/ Gender	Age of sz onset	Types of sz	Interictal SEEG	Ictal SEEG	Response of MAs to initial AEDs	Neuropsychology/ language evaluation	Sz outcome and medications at 12 month follow-up
12/F	3 yrs	MAs, single GTCs	Bursts of gen SWD, independent focal SWD over bilateral temporal regions	Rhythmic SWD at 3 Hz, which are bilateral, synchronous, and symmetrical	Frequency of MAs worsened from 4–5 per day to 80–100 per day while on CBZ and PHB	Normal	In remission on VAL 25 mgm/kg/day and TPA 3.5 mgm/kg/day (LTG did not improve sz outcome)
10/M	4 yrs	MAs alone	Bursts of gen SWD as well as independent focal SWD over bilateral frontal region	Rhythmic SWD at 3 Hz, which are bilateral, synchronous, and symmetrical	Frequency of MAs worsened from 4–5 per day to 80–100 per day while on CBZ and PHB	Normal	In remission on VPA25 mg/kg/day and TPM 3.5 mg/kg/day (LTG did not improve sz outcome)

AED, antiepileptic drugs; CBZ, carbamazepine; EMAs, epilepsy with myoclonic absences; F, female; gen, generalized; GTCs, generalized tonic clonic seizure; LTG, lamotrigine; M, male; MAs, myoclonic absences; PHB, phenobarbitone; SEEG, scalp electroencephalography; SWD, spike wave discharge; sz(s), seizure(s); TPM, topiramate; VPA, valproate; yrs, years.