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Short communication

Do pure absence seizures occur in myoclonic epilepsy of infancy? A case series

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ABSTRACT

Purpose: To assess if absence seizures (ASs) occur in patients with myoclonic epilepsy of infancy (MEI).
Methods: A retrospective chart review was conducted in 37 patients with MEI followed at seven different paediatric epilepsy centres in Italy, between 2002 and 2014. To assess the possible occurrence of pure ASs or absences associated with myoclonias, ASs were defined according to the following criteria: (i) a sudden onset and interruption of ongoing activities; (ii) bilateral polyspikes or spike-and-wave (SW) complexes; spike SW complexes at 2–4 Hz; (iii) duration of AS: 3–30 seconds.
Results: Thirty-seven MEI patients (25 boys and 12 girls) were identified. Nine patients (24.3%) had a history of simple FS during the first year of life. Ten patients (27%) had a family history of epilepsy, and six patients (16.2%) had a family history of FS. In 7/37 (18.9%) patients, during the occurrence of MSs, a total of nineteen brief ASs were captured by video-EEG recordings. ASs occurred both during a brief cluster of rhythmic MSs than after single myoclonic jerks. The ictal EEG abnormalities observed in patients with ASs were similar to the ictal EEG patterns associated with only myoclonias. No differences in relation to gender, family history, ictal EEG discharge were found between patients with myoclonic seizures with ASs and myoclonias without ASs.
Conclusions: Absence seizures can occur in approximately 20% of MEI patients and the occurrence of ASs, though not essential to formulate the diagnosis, do not automatically exclude the diagnosis of MEI.

1. Introduction

Myoclonic epilepsy of infancy (MEI) is probably the earliest form of idiopathic generalized epilepsy (IGE) first described as "benign myoclonic epilepsy of early onset".[1] In 1981, Dravet and Bureau described seven infants with myoclonic seizures and named the syndrome "myoclonic epilepsy in infants".[2] MEI is defined as the occurrence of myoclonic seizures (MS) without other seizure types, except rare simple febrile seizures (FS), in the first 3 years of life. MS are usually very brief (1–3 s), although they may be longer, especially in older children, consisting of pseudo-rhythmically repeated jerks lasting up to several seconds. When MS are repeated, children may be unresponsive, nevertheless, the level of awareness is difficult to assess in isolated MS in this age-related syndrome. Although some authors have reported that absence seizures (ASs) may be associated with myoclonic jerks in MEI patients,[4,5] this issue remains matter of debate.[3] This study was aimed to assess if ASs can occur in patients with MEI.

2. Materials and methods

A retrospective chart review was conducted in 7 pediatric epilepsy centres on patients recruited between January 2002 and

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May 2014. Written informed consent was provided by parents or guardians. All patients met the following inclusion criteria[3]: (1) normal development until seizure onset; (2) seizure onset between 4 months and 4 years of life; (3) myoclonic (including reflex) seizures; (4) electroencephalography (EEG): generalized paroxysms of polyspike (PS) or spike-and-wave (SW) complexes; (5) follow-up of at least 5 years; (6) no evidence of structural or metabolic aetiology.

Patients with myoclonic and atonic seizures were excluded.

To assess the occurrence of pure ASs or absences associated with myoclonias, ASs were defined according to the following criteria: (i) sudden onset and interruption of ongoing activities[6]; (ii) bilateral PS or SW complexes at 2–4 Hz[6]; (iii) duration ranging from 3 to 30 s. All patients underwent sleep and awake video-EEG recordings. Ictal video-EEG recordings were reviewed by all of us in sessions where each seizure was viewed repeatedly and each observer made his/her own comments. Seizure onset, semiological features, frequency, distribution, duration of the seizures, and ictal EEG recordings were analyzed. The pre-ictal state of the patient was recorded as part of the final evaluation (i.e. eyes open or closed, lying or sitting, activity, playing). To evaluate the consciousness involvement, we considered: (1) the presence of upward deviation of the eyes accompanied by the sudden arrest of ongoing activities; (2) interrupted speech accompanied by the arrest of ongoing activities.

Clinical records were reviewed to obtain information including previous FS, first-degree family history of IGE, treatment, and outcome variables. Repeated video-EEGs were performed at the follow-up, after drug withdrawal. Data on school achievements and neuropsychological evaluations were repeatedly obtained during the follow-up.

3. Results

3.1. General features

Thirty-seven MEI patients (25 boys and 12 girls) were identified. The mean and median ages at seizure onset were 22 and 24 months (range 9–36 months). Nine patients (24.3%) had a history of simple FS during the first year of life. Ten patients (27%) had a family history of epilepsy, and six patients (16.2%) had a family history of FS. Physical examination was unremarkable in all patients and none of them had physical dysmorphisms. The main clinical features of the patients are summarized in Table 1.

3.2. Seizure manifestations

At least one ictal polygraphic video-EEG recording was available in all the patients Eighty-nine MSs were recorded. The MSs were predominantly located in the upper limbs and head, with variable intensity in the same child and when comparing children, and from one episode to the next.

In 7/37 (18.9%) patients, during the occurrence of MSs, the ongoing activities were interrupted and alertness was reduced. In particular, in younger patients, ASs were mainly recognized by the occurrence of upward deviation of the eyes accompanied by the arrest of ongoing activities in relation to the onset of the EEG discharge (Fig. 1, see Ref. 7) while in older patients the impairment

Table 1
Clinical features of 37 patients with myoclonic epilepsy of infancy.

Patient	Gender	Family history	Age of onset	Absences	Other seizures	Interictal EEG	Ictal EEG	Therapy	Psychomotor development
1	M	IGE	16	No	None	GSW	GSW/PSW	VPA	NA
2	M	None	14	No	None	PS	PSW	VPA	Normal
3	F	None	21	No	None	Normal	GSW	VPA	NA
4	M	FS	25	No	None	GSW	GSW/PSW	VPA	Normal
5	M	IGE	11	Yes	FS	PS sleep	GSW/PSW	VPA-LEV	Normal
6	M	None	18	No	None	Normal	GSW/PSW	VPA	Normal
7	M	None	25	No	FS	Normal	GSW/PSW	VPA	Normal
8	M	FS	35	Yes	FS	PS	GSW/PSW	VPA	Borderline IQ
9	M	None	30	No	None	Normal	PSW	VPA	Normal
10	F	None	9	No	None	GSW	PSW	VPA	Normal
11	M	None	36	No	None	Normal	GSW/PSW	VPA-LEV	Normal
12	M	IGE	32	No	FS	PS	PSW	VPA	NA
13	F	None	24	No	None	PS	PSW	VPA	Borderline IQ
14	M	None	23	No	None	Normal	GSW/PSW	VPA	NA
15	M	IGE	34	Yes	None	Normal	GSW/PSW	VPA-ETS	Normal
16	M	None	27	No	None	GPSW	GSW/PSW	VPA	NA
17	F	None	10	No	FS	GSW	GSW/PSW	VPA	NA
18	F	IGE	28	No	None	Normal	PSW	VPA	NA
19	M	None	13	No	None	Normal	PSW	VPA	Borderline IQ
20	F	None	14	No	None	GPSW	GSW/PSW	VPA	NA
21	M	None	35	Yes	None	GSW sleep	GSW/PSW	VPA	Normal
22	M	None	13	No	None	Normal	GSW/PSW	VPA	NA
23	F	None	15	No	None	GSW	GSW/PSW	VPA	Normal
24	M	FS	30	Yes	None	GSW awake/sleep	GPSW	VPA	NA
25	M	FS	19	No	None	GPSW	GSW/PSW	VPA	NA
26	M	None	27	No	None	Normal	PSW	VPA	NA
27	F	IGE	14	No	FS	GPSW	GSW/PSW	VPA	Normal
28	M	None	14	No	None	GSW	GSW	VPA	NA
29	F	IGE	17	No	None	Normal	GSW/PSW	VPA	Normal
30	M	None	19	No	None	GPSW	GSW/PSW	VPA	Normal
31	M	IGE	32	Yes	FS	Normal	GSW/PSW	VPA	NA
32	M	None	36	No	FS	GPSW	GSW/PSW	VPA	Normal
33	F	IGE	24	No	None	GSW	GSW/PSW	VPA	NA
34	F	FS	21	No	None	GSW	GSW/PSW	VPA	NA
35	M	None	22	No	None	GSW	GSW/PSW	VPA	Normal
36	F	FS	29	Yes	FS	Normal	PSW	VPA	Normal
37	M	IGE	25	No	None	Normal	GSW/PSW	VPA	NA