Clinical Neurophysiology 125 (2014) 250–254

Contents lists available at ScienceDirect

Clinical Neurophysiology

journal homepage: www.elsevier.com/locate/clinph

Electroencephalographic features of benign adult familial myoclonic epilepsy

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ARTICLE INFO

Article history:
Accepted 1 August 2013
Available online 4 September 2013

Keywords:
Benign adult familial myoclonic epilepsy
Electroencephalography
Generalized spike and wave complexes
Epilepsy with generalized tonic–clonic seizure only

HIGHLIGHTS

- Benign adult familial myoclonic epilepsy (BAFME) is an autosomal dominant disease characterized by infrequent generalized seizures and tremulous myoclonus resembling essential tremor.
- We analyzed the EEG findings of BAFME who were treated at a tertiary referral center.
- Faster frequency of GSW, compared with that in epilepsy with generalized tonic–clonic seizure only (EGTCS), accompanied by PPR may lead to the diagnosis of BAFME.

ABSTRACT

Objective: To investigate electroencephalographic (EEG) features of benign adult familial myoclonic epilepsy (BAFME).
Methods: We reviewed interictal EEG features in patients with BAFME treated between April 2005 and November 2012 at a tertiary referral center. The diagnostic criteria for BAFME were the presence of infrequent generalized tonic–clonic seizures, myoclonus or myoclonic seizures, and autosomal dominant inheritance. Interictal EEG findings of epilepsy with generalized tonic–clonic seizure only (EGTCS) were reviewed for comparison. We randomly selected 10 generalized spike/polyspike and wave complexes (GSW) for each BAFME patient and measured the duration of them. Photic stimulation and hyperventilation were performed in all.
Results: Nineteen (eight men, 11 women) patients with BAFME were included in this study. The mean frequency of GSW was 4.3 ± 1.0 Hz (mean ± SD, $n = 14$) in BAFME and 3.2 ± 0.8 Hz ($n = 10$) in EGTCS. There was a statistically significant difference ($p = 0.008$) between the two. Photoparoxysmal responses (PPR) were noted in 18 (95%) patients with BAFME but 1 (10%) with EGTCS.
Conclusion: Faster frequency of GSW, compared with that in EGTCS, accompanied by PPR may be characteristic EEG features of BAFME.
Significance: These findings may lead the diagnosis of BAFME.

1. Introduction

Benign adult familial myoclonic epilepsy (BAFME) was first reported in Japanese patients by Ikeda et al. (1990). It is an autosomal dominant disease characterized by infrequent generalized seizures and tremulous myoclonus resembling essential tremor. BAFME has been reported from Japan and Europe and has been referred to by various names, such as cortical tremor (Ikeda et al., 1990), BAFME (Yasuda, 1991), familial cortical myoclonic tremor (Terada et al., 1997), familial cortical tremor with epilepsy (Okuma et al., 1997), familial cortical myoclonic tremor with epilepsy (Koelman et al., 2005), familial adult myoclonic epilepsy (Plaster et al., 1999), autosomal dominant cortical myoclonus and epilepsy (Guerrini et al., 2001), and familial cortical tremor myoclonus and epilepsy (Regragui et al., 2006). These various terms represent the same clinical entity.

Previous electrophysiological studies in BAFME have revealed the features of cortical reflex myoclonus, giant somatosensory evoked potentials, enhanced long-loop reflex, and premyoclonus spike detected by the jerk-locked averaging method. On the basis

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1388-2457/$36.00
http://dx.doi.org/10.1016/j.clinph.2013.08.002

of electroencephalographic (EEG) findings, generalized paroxysmal abnormalities and photoparoxysmal response (PPR) have been frequently reported (Guerrini et al., 2001; Striano et al., 2009; Coppola et al., 2011). especially in patients not receiving therapy (Striano et al., 2005). Photomyogenic (formerly called photomyoclonic) response also has been reported (Yasuda, 1991; Striano et al., 2009). Yasuda reported characteristics in 14 patients with BAFME, of whom 10 revealed polyspikes and wave complex, and one revealed spike and wave complex (Yasuda, 1991). Focal temporal or frontotemporal spikes or spike and wave discharges as well as more diffuse abnormalities also have been reported (Guerrini et al., 2001). Ikeda et al. described "numerous small spikes in a multifocal fashion" (Ikeda et al., 1990). When we focused on EEG findings in patients with BAFME treated at a tertiary referral center, we noticed EEG features that lead to the diagnosis of BAFME. In this study, we quantitatively analyzed EEG features of BAFME.

2. Patients and methods

2.1. Patient selections

We used electronic medical records to identify patients with BAFME treated between April 2005 and November 2012 at the University of Occupational and Environmental Health School of Medicine, Japan. The diagnostic criteria for BAFME in this study were the presence of infrequent generalized tonic–clonic seizures, myoclonus or myoclonic seizures, positive family history of epilepsy, and absence of known neurological disorders that cause myoclonic epilepsies. All the patients underwent magnetic resonance imaging to rule out cerebellar atrophy and other structural abnormalities. Because dentatorubral pallidoluysian atrophy is common in the Japanese population and is also associated with seizures, myoclonus or myoclonic seizures, and autosomal dominant inheritance, we distinguished it from BAFME by the absence of cerebellar ataxia, neuroradiographical cerebellar atrophy. All patients underwent electrophysiological evaluation using EEG, surface electromyography, and somatosensory evoked potential tests.

2.2. Electroencephalographic studies

A 21-channel digital EEG recorder (EEG 1100, Nihon Kohden, Tokyo, Japan) was used according to the international 10–20 system of electrode placement, with a sampling frequency of 500 Hz, to make EEG recordings. These recordings were obtained at rest with the eye closed and photic and hyperventilation stimulation, for 30 min in the interictal period. The signals were digitally filtered in the range of 0.53–120 Hz. Frequency of generalized spike and wave complexes or generalized polyspike and wave complexes (GSW) were calculated by measuring the duration (Fig. 1). The amplitudes of the spikes were measured in ear reference montage in the channel where the signal was largest. Spike Amplitude was defined as the magnitude from the peak to bottom (Fig. 1). When multiple EEG recordings were performed in a patient for follow ups, we analyzed the first one. We also investigated antiepileptic drugs and age at the time of EEG was performed. Interictal EEG findings of epilepsy with generalized toni–clonic seizure only (EGTCS) as a subgroup of idiopathic generalized epilepsy (IGE) were reviewed for comparison.

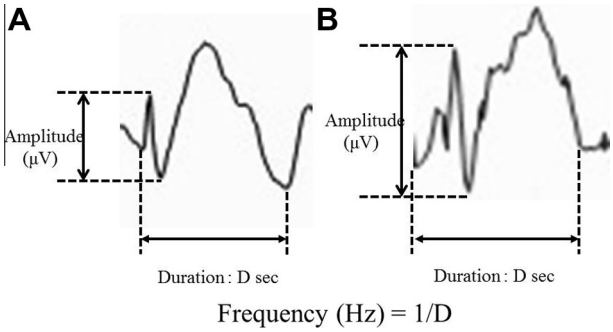

Fig. 1. How to measure the amplitudes of spikes and the duration of generalized spike and wave complexes or generalized polyspike and wave complexes. The reciprocal of the duration is frequency. (A) sample of spike and wave complexes in patient no. 3. (B) sample of polyspike and wave complexes in patient no. 7.

We randomly selected 10 GSW per BAFME patient in the waking stage. The patients with IGE had fewer GSW than those with BAFME; therefore, we selected GSW in the waking stage, whichever was most in the patients with EGTCS who had <10 GSW. As a result 136 generalized spike and wave complexes and four generalized polyspike and wave complexes in the patients with BAFME and 42 generalized spike and wave complexes in the patients with EGTCS were evaluated. We reviewed GSW that were found in the patients at rest with the eyes closed and not during the activation procedure, such as photic stimulation or hyperventilation in the waking stage.

Photic stimulation using stroboscopic diffuse light for 10 s with frequencies of 3, 6, 9, 12, 15, and 20 Hz for was performed.

Table 1
Frequency of generalized spike/polyspike and wave complexes (GSW) and amplitudes of spikes in BAFME.

patient no./gender	age at Nov. 2012 (years)	age at EEG performed (years)	antiepileptic drugs (mg/day)	Interictal EEG findings		
				frequency of GSW	amplitudes of spikes	PPR
1/M	77	75	CZP 2	4.2 Hz	81.9 μV	+
2/F	49	47	PHT 200	4.8 Hz	163.0 μV	+
3/F	57	55	–	5.4 Hz	94.5 μV	+
4/F	55	54	CZP 0.5	6.6 Hz	18.8 μV	+
5/F	73	67	CZP 1.5	3.6 Hz	60.8 μV	+
6/F	80	76	CZP 1.5	4.9 Hz	75.3 μV	+
7/M	56	55	PHT 250/PB 60	2.9 Hz	66.0 μV	+
8/F	49	48	CZP 1.5	5.2 Hz	47.4 μV	+
9/F	69	69	–	4.6 Hz	128.1 μV	+
10/M	75	68	PHT 100	4.2 Hz	69.4 μV	+
11/M	79	73	PHT 300 /PB 8/TMO 1,000	2.9 Hz	108.8 μV	+
12/M	71	70	–	3.7 Hz	85.8 μV	+
13/F	43	43	ZNS 200	3.3 Hz	40.2 μV	+
14/F	64	64	PHT 200/VPA 600	4.4 Hz	81.6 μV	+
Mean	64	62		4.3 Hz	80.1 μV	100%

EEG, electroencephalography; GSW, generalized spike and wave complexes or generalized spike and wave complexes, CZP, clonazepam; PHT, phenytoin; PB, phenobarbital; TMO, trimethadione, ZNS, zonisamide; VPA, valproate.