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Absence status epilepticus in monozygotic twins with Jeavons syndrome

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ABSTRACT – As a generalized form of nonconvulsive status epilepticus (NCSE), absence status epilepticus is the most common form. It manifests as prolonged, confusional states of varying severity, and continuous or repetitive generalized discharges of spikes, multiple spikes, and slow waves on EEG. Jeavons syndrome (JS) is a new type of epilepsy syndrome. Hitherto, only four sets of monozygotic twin with JS have been reported. Absence status epilepticus occurring in monozygotic twins with JS have not been reported. Here we report on monozygotic male twins of Chinese origin with JS. Both of them presented with status epilepticus with eyelid myoclonia and absences. *[Published with video sequences]*

Key words: monozygotic twins, Jeavons syndrome

Nonconvulsive status epilepticus (NCSE) accounts for approximately one-quarter of all cases of status epilepticus (Cascino 1993). As a generalized form of NCSE, absence status is the commonest form, ranging from 53 to 94% in different studies (Tomson *et al.* 1992, Shorvon 1995). It is characterized by prolonged confusional states of varying severity and continuous or repetitive generalized discharges of spikes, multiple spikes, and slow waves on EEG. Recently, new epilepsy syndromes commonly seen in ASE have been reported. Among these, epilepsy with eyelid myoclonia and absences (Jeavons syndrome) was included. Jeavons syndrome (JS) was first described by Jeavons (1977). JS is characterized by eyelid myoclonia with and without absences, eye closure-induced seizures, photosensitivity, unique clinical and EEG features and often genetic clustering (Panayiotopoulos 2005). JS is generally considered to be a rare condition. Its prevalence is around 2.7% among adult epileptic disorders, 12.9% among idiopathic generalized epilepsies (IGE) with typical absences (Giannakodimos and Panayiotopoulos 1996), and with an unknown prevalence in children. Hitherto, there have been only four reports of JS in monozygotic twins (Striano *et al.* 2002, Masao *et al.* 2005, DeMarco 1989). However, no reports of ASE in monozygotic twins have been presented. Herein, we report a case of monozygotic male twins of Chinese origin

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with JS, in whom ASE developed. We have followed up these monozygotic twins for two years.

Case reports

The monozygotic twins were male, aged 13 years. There was no family history of epilepsy and no consanguinity. No abnormal events had occurred during the pregnancy, neonatal period, or infancy. However, at the age of eight, eyelid myoclonia with occasional episodes of upward eyeball deviation was noticed in both children. Eyelid myoclonia was paroxysmal and its duration ranged from several minutes to hours. Most eyelid myoclonia occurred after eye closure and worsened in bright light. When myoclonia worsened, brief lapses in concentration occurred. At ten years of age, twin 1 displayed status epilepticus with eyelid myoclonia and absence lasting more than five hours. Despite subsequent improvement, there was persistent eyelid myoclonia. The EEG in the local hospital showed paroxysmal bilateral 2.5-4.0 Hz generalized polyspikes and slow waves, with middle-high amplitude and frontal-temple lobe predominance. The brain CT was normal. Although the diagnosis was not confirmed, phenobarbitone was prescribed (30 mg per day), which improved twin 1's clinical symptoms to some extent. At the age of 12, twin 1 presented with generalized tonic clonic seizures (GTCS) without any obvious predisposing factors. He was then referred to our hospital where the EEG showed continuous bilateral 2-4.0 Hz generalized polyspikes and slow waves with high amplitude. Twin 2 was similar to twin 1, but the severity was less, and he had no GTCS. His EEG was similar to that of his brother. Absence seizures were diagnosed and sodium valproate (VPA) was prescribed (250 mg per day) for both twins. During the followed-up period of more than one year's treatment with VPA, the clinical symptoms in both twins were remarkably improved and were without any effect on normal life, although uncontrollable eye blinking and brief absence occurred occasionally. Over the past 20 days, VPA was discontinued by the twins and the clinical symptoms worsened, especially on awakening. The twins returned to our hospital, underwent cerebral MRI, which was normal for both twins, and video-EEG *(figures 1, 2)* were performed. The diagnosis of Jeavons syndrome was confirmed. During the video-EEG, status epilepticus with eyelid myoclonia and absences occurred in both boys *(see video sequence)*. The patients could not identify their parents, distinguish right from left, perform simple calculations, and there was slowness of judgment and planned movements. This lasted for fourhours, 50 minutes and two hours, 45 minutes, in twin 1 and twin 2 respectively.

The video-EEG demonstrated continuous, generalized, high to very high amplitude irregular polyspike and spike-wave discharges and blinking artifacts shown before trains of spike-wave discharges. These then resolved spontaneously. Sodium valproate was continued (250 mg bid/day). The patients' symptoms improved without GTCS or absence status epilepticus occurring again, but brief episodes of eyelid myoclonia with the occasional absence

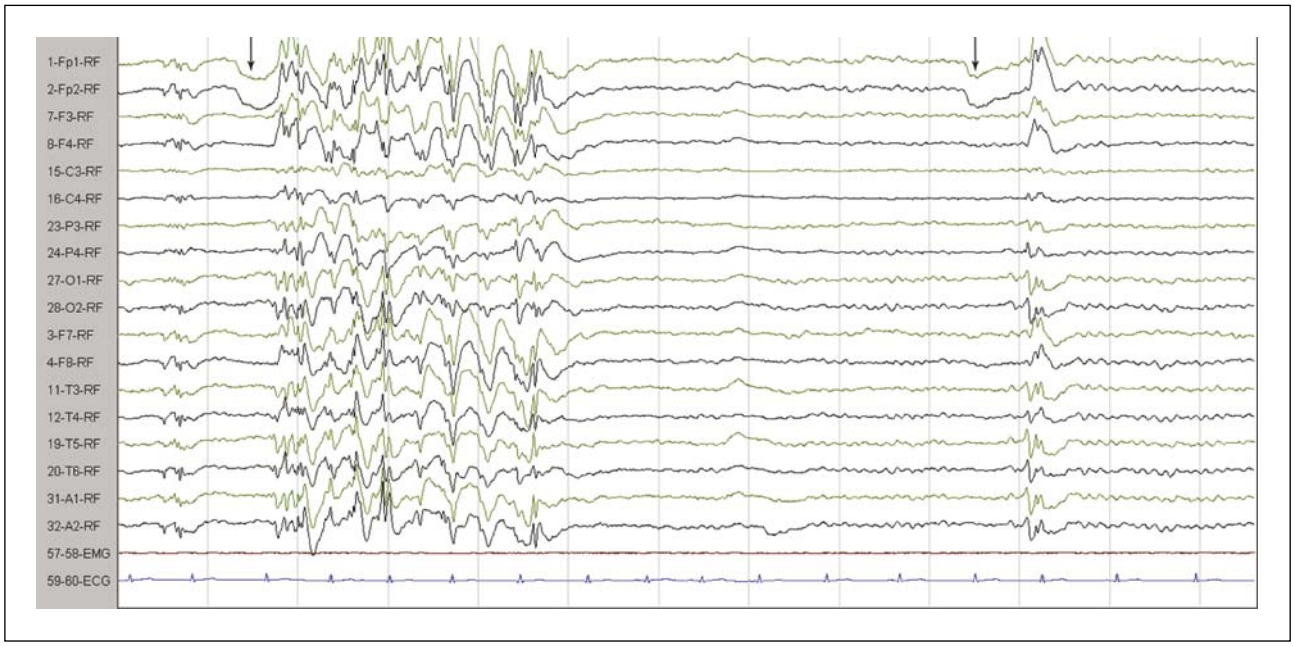

Figure 1. The patient (twin 2) was in a conscious, alert, resting state. The clinical manifestation was paroxysmal, fast, repetitive jerks of the eyelids. Video-EEG showed main α background activity, bilateral frequent high amplitude 2.5-6HZ GSWD and sharp and slow wave complexes, which occurred immediately within 0.5-2s after eye closure (arrow) and briefly.

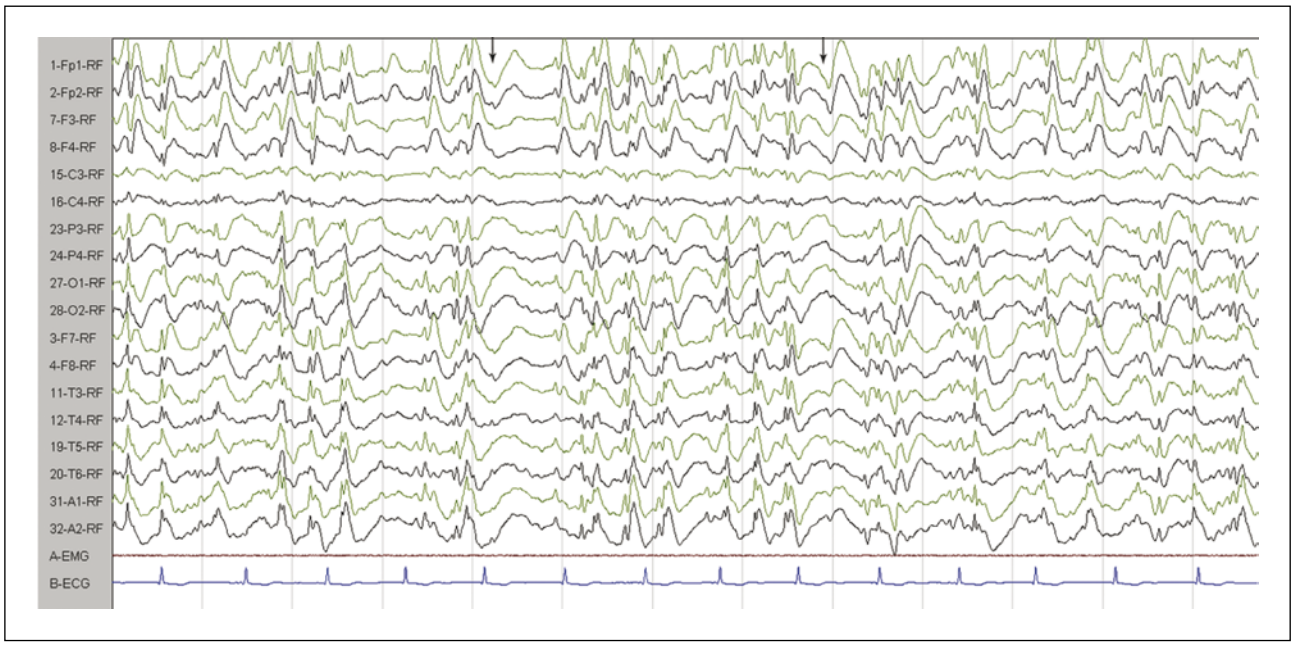

Figure 2. Status epilepticus of EMA (twin 1). The clinical manifestation was continuous, fast, arrhythmic jerks of the eyelids, often associated with retropulsion and repetitive brief lapses of consciousness. The patients could not identify their parents, distinguish right from left or perform simple calculations, and there was slowness of judgment and planned movements. This lasted for four hours, 50 minutes and two hours, 45 minutes in twin 1 and twin 2, respectively. The video-EEG demonstrated continuous, generalized, high to very high amplitude, irregular polyspike and spike-wave discharges and blinking artifacts (arrow), shown before trains of spike-wave discharges.

occurred when emerging into bright light. Their neurological and physical examinations were normal, and their academic performance remains within the average range.

Discussion

ASE is a common and often undiagnosed condition in IGE, which requires appropriate attention and treatment. It has been reported that typical absence status occurs in varying rates ranging from 20 to 38% patients with IGE and typical absences (Michelucci *et al.*1996, Panayiotopoulos *et al.* 1992, Wolf and Inoue 1984). Agathonikou *et al.* reported that typical absence status was syndrome-related, and the frequency of ASE varied considerably with different IGE syndromes. The frequency of ASE in eyelid myoclonia with absences (EMA) is much lower than that in perioral myoclonia with absences or phantom absences with GTCS. Almost half of patients with perioral myoclonia with absences or phantom absences with GTCS will experience ASE (Agathonikou *et al.* 1998). ASE was found to occur with a frequency of 1.2 per 100 cases annually in juvenile myoclonic epilepsy (Dziewas *et al.* 2002). The low prevalence of ASE in juvenile myoclonic epilepsy is consistent with previous reports (Kimura and Kobayashi 1996, Reutens and Berkovic 1995). ASE is considered to be rare in the core syndrome of childhood absence epilepsy. However, ASE is common in Doose syndrome (myoclonic-astatic epilepsy). In juvenile absence epilepsy, ASE is relatively uncommon, although varied prevalence has been reported, which may be due to the different diagnostic criteria for juvenile absence epilepsy.

ASE in EMA is always situational and the episodes of ASE are typically precipitated by factors such as menstruation, hypoglycaemia, hyperventilation, flashing or bright lights, sleep deprivation, or as in our patients, it may be mainly a result of discontinuation of anti-convulsant medication. The ASE in our patients resolved spontaneously, unlike as in other syndromes such as perioral myoclonia with absences and phantom absences, which frequently ends with GTCS. These other syndromes should be terminated with immediate, self-administered medication of oral midazolam or rectal benzodiazepines (Agathonikou *et al.* 1998, Baykan *et al.* 2002). ASE in our patients persisted for several hours, which is consistent with other reports, in which the duration of ASE varied from hours to days. (Masao *et al.* 2005).

Most reports of status epilepticus (SE) with EMA have described an absence status, as in our patients. The clinical manifestation was continuous eyelid myoclonia and repetitive, brief lapses of consciousness. The patient could not identify their parents, distinguish right from left, perform simple calculations or planned movements, and had slowness of judgment. The EEG typically demonstrates continuous, generalized, high-very high amplitude, irregular 2.5-to 6-HZ polyspike and spike-wave discharges (Wakamoto *et al.* 1999). In addition, eyelid myoclonia

Legend for video sequence

The clinical manifestation was continuous, fast, arrhythmic jerks of the eyelids, often associated with retropulsion and repetitive, brief lapses of consciousness. The patients could not identify their parents, distinguish right from left or perform simple calculations, and there was slowness of judgment and planned movements. This lasted for four hours, 50 minutes and two hours, 45 minutes, in twin 1 and twin 2, respectively.

with absences occurred with polyspikes and 2.5-6 Hz polyspike-slow waves; however, without absences it occurred with mainly polyspike discharges of brief (1-2 s) duration (Giannakodimos and Panayiotopoulos 1996). In another report, however, long-lasting eyelid myoclonia presented with only extremely brief, impairment of consciousness. There were no differences on the EEG between eyelid myoclonia with and without absences, and absences were extremely brief and mild, allowing the patient to perform his daily activities well in spite of severely persistent eyelid myoclonia. There were continuous 2.5-4.0 Hz polyspikes and/or slow wave bursts on EEG while experiencing SE (Masao *et al.* 2005). This may indicate that there are different types of SE with EMA, such as eyelid myoclonic status and absence status. Additionally, in our patients, video-EEG during ASE showed continuous, generalized, high-very high amplitude, irregular 2.5- to 6-HZ polyspike and spike-wave discharges, different from that of the phantom absences, which showed continuous, generalized, mainly 3-Hz spike/multiple spike slow wave activity. It was also different from that of perioral myoclonia with absences during ASE, which showed continuous generalized polyspikes and slow-wave complexes at 2.5-4 Hz, accompanied by perioral twitching.

In summary, this is the first report of ASE occurring in both monozygotic twins. In our genetically identical twins, there are some variations in the phenotype. This may be due to environmental factors (Briellmann *et al.* 2001). Genetic factors may play an important role in JS. These genetic influences however, are unclear. So linkage analysis or other genetic approaches may be helpful in elucidating the basis for the genetic clustering phenomenon in JS. □

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