Atypical focal ESES/CSWSS: a clinical case

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Abstract

We report an adolescent female showing the onset, at the age of 16, of tonic-clonic and focal motor seizures. The sleep EEGs disclosed continuous Spike Wave discharges occupying more than 85% of non REM sleep focalized over the left fronto-temporal regions and persisting for almost 2 years. The epileptic seizures were ultimately controlled by valproate and clobazam and the patient did not develop any cognitive impairment. Moreover 1.5 T cerebral MRI and FDG PET were normal. We suggest that atypical cases of ESES with focal distribution, onset during adolescence and absence of cognitive impairment may be observed and need prompt recognition.

KEY WORDS: sleep, epilepsy, encephalopathy.

Introduction

Encephalopathy with status epilepticus in sleep is defined as an age-related and self-limited disorder characterized by peculiar electro-clinical features: neuropsychological impairment with global or selective regression of cognitive or expressive functions; motor impairment, in the form of ataxia, dyspraxia, dystonia or unilateral deficit; epilepsy, with focal and generalized seizures (unilateral or bilateral clonic seizures, tonic-clonic seizures, absences, partial motor seizures, complex partial seizures or epileptic falls) and typical EEG findings, i.e. electrical status epilepticus in sleep (ESES) or Continuous Spikes and Waves During Slow Sleep (CSWSS). ESES is characterized by nearly continuous spike-wave (SW) discharges in slow wave sleep, usually with a frequency of 1.5-3 Hz, with bilateral distribution, first described in 1971 by Patry, Lyagoubi and Tassinari (1). This EEG pattern must be present for at least the 85% of slow wave sleep and persist for 3 or more recordings over a period of at least 1 month (2), even if a SW index greater than 85% is no longer required by the International League Against Epilepsy to fulfill the diagnostic criteria for ESES (3).

During wakefulness, the EEG is usually abnormal, often showing paroxysmal foci over the frontotemporal or centrotemporal regions or brief bursts of more diffuse SW activity (4). However, in non-rapid eye movement (NREM) sleep, the discharges are significantly activated and become nearly continuous. Even if these discharges arise focally in one hemisphere, they then rapidly propagate within and between hemispheres, thus appearing spatially diffuse (4).

The age of onset is variable, ranging from 1 to 14 years, with a mean onset between 4 and 8 years (5), however few cases have been reported with an adult onset (6).

The etiology is unknown, a correlation with structural brain abnormalities is frequently described. In fact Magnetic Resonance Imaging (MRI) abnormalities are commonly found such as cortical dysplasia, congenital stroke, diffuse atrophy, white-matter changes, abnormal or delayed myelination, tubers and Chiari II malformation (7, 8).

The pathophysiological mechanisms underlying the appearance of cognitive and behavioral disorders in ESES are still poorly elucidated. It has been hypothesized that epileptic EEG paroxysms may interfere with physiological functions and, possibly, with neuroplasticity processes involved in higher cortical functions (such as learning and memory) that occur during slow wave sleep (9). The duration of ESES and the localization of interictal activity may be responsible for the degree and the type of cognitive dysfunction (9).

ESES/CSWSS is a self-limited EEG pattern that usually disappears within 3-4 years with a contemporaneous disappearance of seizures in almost all cases. However, despite the disappearance of the EEG...
abnormalities and seizures, the prognosis is not always good due to the usual persistence of neuropsychological impairment (7).

Aim of the treatment is to control seizures and to improve neuropsychological function, which requires a normalization of the encephalographic pattern. Traditional antiepileptic drugs play only a minimal role in controlling this syndrome (4). Carbamazepine, phenytoin and barbiturates may reduce seizures, but may worsen the neuropsychological outcome and EEG discharges. Valproic acid, ethosuximide, levetiracetam and benzodiazepines may be effective, as reported in a few small case series (10, 11).

High-dose benzodiazepines and steroid therapy are most commonly used showing more benefits than traditional antiepileptic drugs (AED). Other therapies, including intravenous gamma globulin, the ketogenic diet, and surgical treatment, have shown efficacy in some cases.

The aim of this paper is to describe a patient with a late-onset focal ESES/CSWSS without cognitive impairment and good response to AED after three years of follow-up. The description of this atypical case may contribute to widen the clinical spectrum of this condition.

Clinical description

We describe the case of a girl who came to our attention at the age of 16. In her family history she reported epilepsy in a cousin and arousal disorders in her father and older sister. Clinical information about pregnancy, delivery and psychomotor development were unremarkable. At the age of 16, during afternoon naps and night-time sleep, she presented three tonic-clonic seizures. She was admitted to our department and underwent various EEG recordings, neuropsychological assessment and neuroradiological evaluation.

Electroencephalographic recordings during wakefulness showed a normal background activity and frequent, brief SW discharges over the left fronto-temporal regions (Fig. 1), spreading over the adjacent regions, especially during hyperventilation. Intermittent photic stimulation was ineffective. A video-polysomnographic recording showed an abnormal organization of both NREM and REM sleep stages, with the physiological sleep patterns only barely recognisable. During NREM sleep, we recorded subcontinuous spike and slow wave discharges over the left fronto-temporal regions, with sporadic contralateral diffusion, lasting for more than 85% of the total amount of NREM sleep (Fig. 2). Furthermore, the patient presented stereotyped seizures with right hand-nose scrubbing, chewing and dystonic postures of the right arm. During these episodes, the EEG showed a brief desynchronization of cerebral activity, followed by brief, diffuse theta-delta rhythmic discharges with subsequent rapid and rhythmic activity over the frontal regions, bilaterally.

Figure 1 - EEG recording during wakefulness showing sporadic spike and waves over the left fronto-temporal region. LF: 1.6 Hz; HF: 30 Hz; sweep 200; sens: 100 µV/cm.
We recorded this EEG pattern during different follow-up video-eeg over two years. She underwent cognitive level assessment that did not disclose any abnormality, as well as 1.5 Tesla brain MRI and 18-FDG PET which were normal. She was initially treated with oxcarbazepine (OXC) with clinical improvement for almost one year. The subsequent attempt of increasing the dosage of OXC induced the appearance of vertigo, dizziness and sleepiness and therefore OXC was gradually shifted to valproic acid (VPA), up to the dosage of 800 mg/day, and clobazam (CLB) at the dose of 10 mg/day with disappearance of seizures and no adverse effects. Her last EEG recording, performed at the age of 19, showed only sporadic sharp waves over the left fronto-temporal derivations during wakefulness and sleep (Fig. 3). She was seizure-free and her cognitive level was unchanged.

Conclusions

Encephalopathy with status epilepticus in sleep is a condition characterized by typical EEG findings (ESES/CSWSS), seizures and cognitive and motor impairment of various degrees. It is generally age-related and self-limited but usually severe neuropsychological deficits persist (1). The EEG abnormalities are usually diffuse and, until now, only few cases have been described with a focal localization of ESES. In these cases structural abnormalities, such as polymicrogyria (PMG), shunted hydrocephalus, porencephalic lesions associated with PMG and thalamic lesions, have been found (12). Furthermore selective cognitive deficits associated with the cerebral areas showing the predominat SWs localization have been reported as distinctive features of focal ESES (2, 9).

Our case represents an atypical presentation of focal ESES/CSWSS. In fact the presence of focal SW discharges for more than 85% of NREM sleep, persisting over several EEG recordings, supports the diagnosis of ESES/CSWSS. However the age of onset, the absence of cognitive and motor deterioration and a normal MRI represent atypical features, never described until now. Nevertheless, in order to rule out the presence of a not recognized focal lesion, a 3 Tesla MRI should be performed. Even if some Authors reported a negative response to some AEDs with worsening of the electroclinical features (12), she was initially treated with OXC with clinical improvement for almost one year. Moreover, despite the persistence of the ESES pattern, the patient showed a good clinical response to the subsequent antiepileptic treatment (VPA and CLB) with total recovery after the age of 18. We suggest that focal ESES/CSWS should be taken into account by clinicians also in adult patients without cognitive impairment but our case report deserves further confirmation.
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Figure 3 - EEG recording during NREM sleep after 3 years of follow-up. LF: 1.6 Hz; HF: 30 Hz; sweep 20''; sens: 100 µV/cm.

References