

Identical Twin Discordant for Electrical Status Epilepticus of Sleep (ESES)

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Introduction

Electrical status epilepticus in sleep (ESES), also known as Continuous Spike Wave during sleep (CSWSS), is a rare phenomenon, occurring in less than 1% of children with epilepsy [1].

While some experts use the terms interchangeably, other refer to ESES strictly as the electrographic pattern of frontally dominant spike wave discharges that comprise 85% of non-REM sleep, while CSWSS comprise this EEG pattern, together with the clinical syndrome of regression and epilepsy. The discovery of ESES is considered to be a neurological emergency as permanent intellectual deterioration is associated with the ESES EEG pattern, when refractory. Age of onset has been reported between 3-14 years. The EEG pattern of sleep-induced generalized spike-and wave discharges (1.5-3.5 Hz), occurring continuously or discontinuously, and lasts for months to years. The International League against Epilepsy (ILAE) has determined that ESES is neither a focal nor generalized syndrome [1]. Indeed, this condition has been determined symptomatic, idiopathic (presumed genetic), or cryptogenic (unknown).

In the symptomatic group of patients with ESES, an underlying brain structure can be determined for approximately 1/3 of patients [2]. Polymicrogyria is the most frequent cause [3]. Other structural causes include stroke, particularly involving the thalamus [4]. Indeed, one theory suggests that early life injury to the thalamocortical pathways induces a secondary bilateral synchrony that generates ESES in these symptomatic patients [4].

Surgery to remove the lesion has been effective in reducing seizures and improving EEG and neuropsychological function in children with such focal lesions [4].

Less is known about idiopathic/cryptogenic causes of ESES, making up about 1/3 of all cases [2].

One theory is that ESES falls along the continuum of three conditions: benign partial epilepsy of childhood, acquired aphasia (Landau-Kleffner syndrome) and ESES. The benign partial epilepsy of childhood, including benign rolandic epilepsy (BRE) is considered at the "mild" end of the continuum with ESES, associated with intellectual deficits, considered at the severe end.

The intellectual deterioration in ESES has been attributed to the frontal lobe localization (subserving judgment and executive function), in comparison to the left temporal (underlying speech) distribution of repetitive epileptiform discharges in Landau-Kleffner syndrome. Similar to BRE, the idiopathic/cryptogenic forms often remit in adolescence, with an eventual normalization of the EEG. However, unlike BRE, there is often a permanent intellectual impairment if before cessation, the patient's EEG pattern shows unremitting ESES. BRE has long thought to run in families, however recent twin data counters this [5] (discussed below). Still, genetic BRE studies have demonstrated linkage of centrotemporal sharp wave EEG to 11p13 and several polymorphic markers in the Elongator Protein Complex [4] gene

(ELP4) as reviewed by Rudolf et al. [6] Further, genetic factors have been hypothesized to underlie ESES [6].

The following genes have been found in association with CSWSS: CHRNA7, PCYT1B, neuroserpin, 8q 12.3q 13.2 microdeletion, DOK5 on chromosome 20q13, as reviewed by Sanchez et al [7].

There has only been one report of ESES in monozygotic twins: they were concordant for ESES [8].

The following is a description of monozygotic 5 year old twins, discordant for ESES.

Case

The patient is a developmentally normal, identical twin who was born nearly full term without complication. At age two, the patient presented with occasional seizures in the form of nocturnal focal seizures affecting her face. Seizures increased in frequency so that by age four she was placed on oxcarbazepine, but this was discontinued due to behavioral side effects. Initially, MRI was normal and EEG showed bicentral epileptiform discharges that were of high amplitude associated with sleep. This was interpreted as being consistent with BRE. Within one year, however she was having focal, atypical absence, and generalized seizures refractory to valproic acid, lamotrigine, and clonazepam. EEG at that time showed continuous spike wave discharges associated with non-REM sleep. Two years later, besides having medically refractory seizures, she showed significantly reduced function: she could not walk, talk, or understand directions. Follow-up MRI after one year showed mild white matter volume loss.

Pet study identified decreased activity in the bilateral temporal lobes. At this point, it was decided to initiate oral steroids (hydrocortisone) and her clinical seizures dramatically improved within one month. The EEG, however, continued to show ESES. After the administration of diazepam at bedtime, the EEG improved. Nine months after beginning hydrocortisone, the girl was virtually seizure free and regained speech and mobility. She was functioning normally in kindergarten. After a slow wean of hydrocortisone, however, seizures increased and cognitive disturbance returned. A longer course of prednisone was begun and she has improved mentation and seizure control. The patient's identical twin has not experienced seizures; an EEG obtained on this sister in the awake and sleep states was normal.

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Discussion

The fact that our patient's identical twin showed no abnormality supports an environmental trigger or epigenetic cause of ESES rather than the genetic cause that has been suggested [1,5].

Again, this report is in contrast to the only other report of ESES in monozygotic twins, showing concordance [8]. Epigenetics likely accounts for the concordance rate of 75%, instead of 100% seen in epilepsy identical twin studies, e.g. Vadlamudi et al. [9] However, large twin studies have shown high concordance rates for generalized epilepsies, and febrile seizures, more than partial seizures [9,10]. ESES was not included in these twin studies. Further, the recent surprising finding of an absence of concordant twin pairs in a study of 18 twins of whom at least one had classic BRE, suggests that non-inherited factors are of major importance in BRE5, believed to be a kin to ESES. Similarly, our case supports environmental/epigenetic factors underlying ESES.

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