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SLEEP MEDICINE PEARLS

A Sleepy Patient With Frequent Falls

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A 25-year-old man was referred to our sleep center with a suspected diagnosis of narcolepsy with cataplexy. He complained for several years of excessive daytime sleepiness. He described "cataplexy-like" events manifest as his arms and legs "twitching" prior to his dropping to the ground with consciousness preserved. This predominantly occurred in early mornings soon after waking up. Given a presumptive diagnosis of narcolepsy with cataplexy he was empirically prescribed armodafinil by his physician.

On further questioning he endorsed "twitches" consisting of bilateral jerks of his upper and lower extremities that clustered about an hour after his waking up from sleep, sometimes resulting in loss of balance and falls. He denied any association with positive or negative emotions. His mother had similar complains as a child but was never treated until she experienced generalized tonic-clonic seizures as an adult. His wife reported that he also snores loudly in sleep. Physical examination showed body mass index of 28 kg/m^2 and Mallampati grade IV upper airway. Polysomnography demonstrated an apnea-hypopnea index of 17 events/h and frequent generalized spike-wave discharges (**Figure 1** and **Figure 2**).

Question: What is the diagnosis?

Figure 1—10-second epoch of polysomnography showing generalized polyspike and wave discharge in sleep.

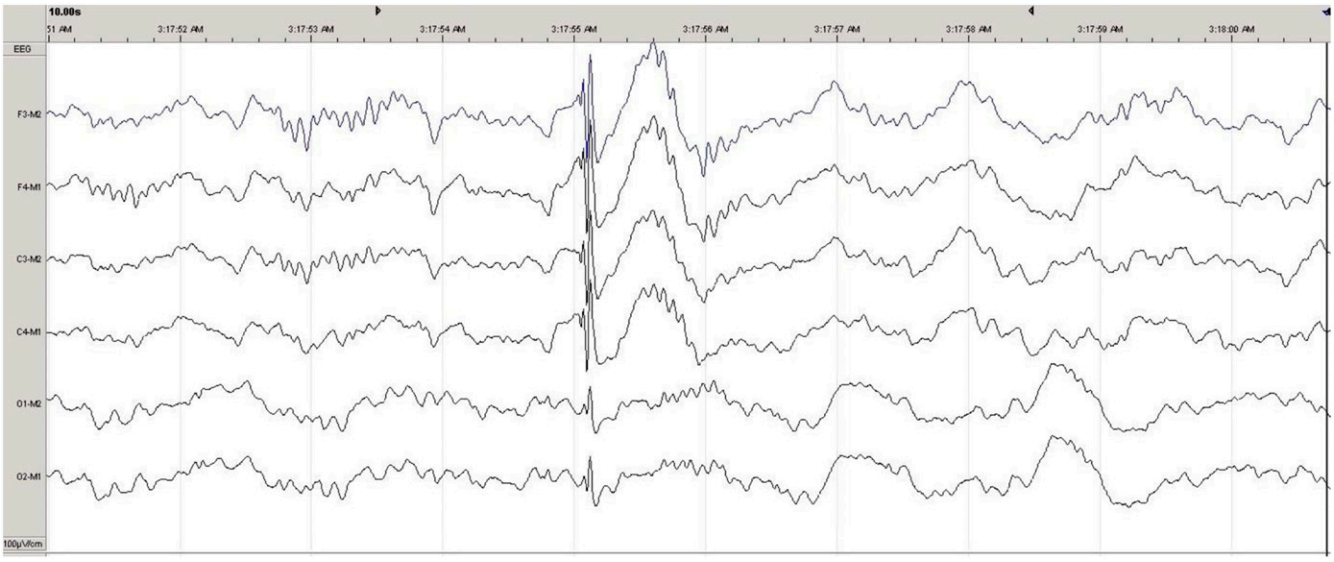
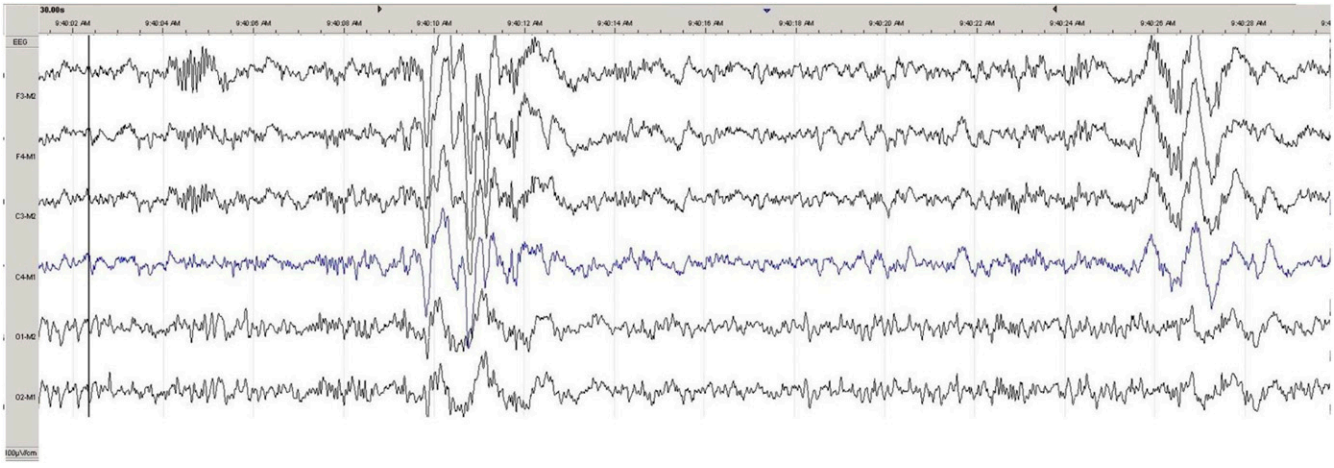


Figure 2—30-second epoch of polysomnography showing generalized polyspike and wave discharge in sleep.



Answer: Juvenile myoclonic epilepsy (JME) with obstructive sleep apnea (OSA)

DISCUSSION

JME syndrome associated with sleep usually manifests between 8 and 26 years of age. The key feature of JME is myoclonic jerks without loss of consciousness that typically occur shortly after awakening. They are brief, single or repetitive, and often occur in brief clusters. The upper extremities are commonly affected resulting in patients dropping objects from their hands.[1] Myoclonic jerks may involve the lower extremities, resulting in flexion of the knees and falls, as present in our patient. Generalized tonic-clonic and absence seizures may also be observed. Seizures are typically precipitated by sleep deprivation and alcohol use.[2] Electroencephalography results showed generalized four to six per second spike/polyspike and wave discharges. Although JME is a chronic condition, antiepileptic drugs, particularly valproic acid, are very effective in controlling seizures. Levetiracetam and lamotrigine are alternative options in women of childbearing age given teratogenicity risk with valproic acid use.[3] The diagnosis of JME is often missed, resulting in delay in appropriate treatment.[4] In our patient, myoclonic jerks resulting in falls were misinterpreted as cataplexy. His excessive daytime sleepiness (prompting a misdiagnosis of narcolepsy) was likely due to untreated OSA. OSA is twice as common in adults with epilepsy than in age-matched controls, and the incidence increases with age.[5] Managing OSA with positive airway pressure has been shown to improve seizure control.[6] Most nocturnal seizures can also cause significant disruption of sleep architecture, resulting in poor sleep efficiency and excessive daytime sleepiness.[7] Our patient was started on levetiracetam for seizures and continuous positive airway pressure therapy for sleep apnea. With this, he had

complete resolution of his myoclonic jerks and daytime sleepiness. Lifestyle modifications should include avoidance of alcohol and maintaining a regular sleep-wake cycle. Sleep clinicians must familiarize themselves with clinical features of various sleep-related epilepsies in order to prevent diagnostic delay and to initiate appropriate treatment.

SLEEP MEDICINE PEARLS

1. JME is a sleep-related epilepsy syndrome characterized by myoclonic jerks that occur upon awakening from sleep, often triggered by sleep deprivation and alcohol use.
2. Obtaining good history and identifying clinical and electroencephalographic features is key to rendering a correct diagnosis.
3. Electroencephalographic features include generalized four to six per second spike/polyspike and wave discharges.
4. Antiepileptic drugs along with proper sleep hygiene are very effective in controlling seizures in JME.

CITATION

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ABBREVIATIONS

JME, juvenile myoclonic epilepsy
OSA, obstructive sleep apnea

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DISCLOSURE STATEMENT

All authors have read and approved this manuscript. The authors report no conflicts of interest.