Chapter 7
Juvenile Myoclonic Epilepsy

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Case Presentation

A 21-year-old right-handed white female with well-controlled epilepsy developed normally without medical conditions or risk factors. Seizure onset was noted at 17 years of age after a night of "cramming" for a final examination in history class. After the test she stayed out with friends until 1 AM. She admitted to drinking four Red Bull® energy drinks and staying out late with friends before returning home to sleep. The following day she experienced headache and nausea. When she went to brush her teeth, her right arm jerked, and her toothbrush was jettisoned from her hand. She went to eat breakfast but continued to be "shaky" and found it difficult to eat her cereal due to jerky motions that created trouble guiding the spoon to her mouth. She then described the occurrence of similar jerking in the morning in the first half hour of awakening over the last 2 years. As she was telling her parents about the night before, she suddenly turned her head to the left, let out a scream, and fell to the ground. She was unconsciousness and manifested generalized tonic stiffness and clonic jerking bilaterally for 1 min. She was then tired, sleepy, and confused. Her parents called 911, and she was taken to the ED. In the ED, she was disoriented and confused but without focal or lateralizing features to her neurological examination. A CT brain was normal. An EKG and laboratory testing included a normal CBC, liver function studies, and electrolyte panel, and creatinine was normal. She was given intravenous levetiracetam and was admitted to the hospital overnight. An MRI of the brain was normal and an EEG had the following results (see Fig. 7.1).

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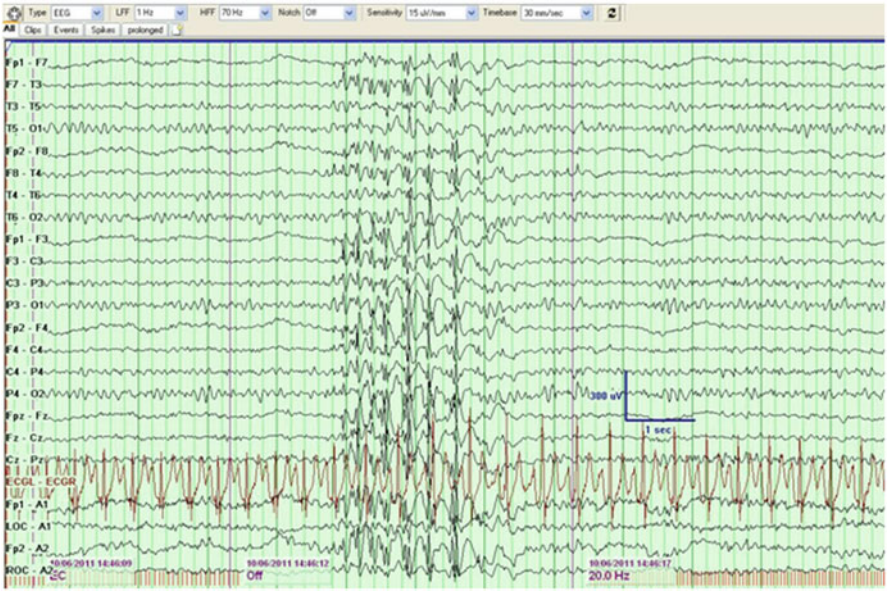

Fig. 7.1 Generalized spike- and polyspike-and-waves with a repetition rate of 4–5 Hz at the onset of a 2-s burst of interictal epileptiform activity that occurred without clinical signs

Clinical Questions

1. Does this patient have epilepsy?
2. What would you expect the EEG to demonstrate?
3. What clinical features characterize this epilepsy syndrome?
4. What is the best treatment for this condition?
5. What is the prognosis for this patient?

Diagnostic Discussion

1. Our patient had only a single generalized tonic-clonic seizure; however, generalized upper-body predominant single lightening-like jerks that occur in the morning and result in spilling drinks or throwing objects such as a toothbrush represent myoclonic seizures. These are brief shock-like jerks that involve the shoulders, face, arms, and legs and may precede the onset of convulsions by 2 years. They appeared in our patient at 15 years of age, the peak age of onset. The clinical presentation of juvenile myoclonic epilepsy (JME) is an initial GTC seizure that occurs in otherwise healthy individuals the morning after a night of sleep deprivation and/or alcohol consumption. The peak age of onset is typically 12–18 years of age. In this case, the seizures were precipitated by sleep deprivation and

the use of energy drinks. Sleep deprivation, alcohol, stimulant drugs, strobe lights or video games, and energy drinks may all act as precipitating factors.

2. EEG supports a clinical diagnosis of epilepsy. In this case, an interictal EEG demonstrated generalized spike- and polyspike-and-wave at >3 Hz. The interictal EEG is abnormal in 50–85 % of untreated patients with JME. The characteristic electrographic feature includes "fast" 3–5 Hz generalized bilateral frontocentral predominant, symmetric synchronous polyspike-and-waves as in the case above. Photosensitivity seen with photic stimulation occurs in 30–50 % of patients stimulated at midrange frequencies (about 15 Hz). This supports a generalized mechanism in a patient with a clinical diagnosis of seizures. The polyspike formation is suggestive of myoclonic seizures.
3. JME is the most common form of genetic generalized epilepsies and the most common cause of primary GTC seizures. It is characterized by adolescent-onset myoclonic jerks that occur with morning predominance. Generalized tonic-clonic seizures are seen in 95 % of patients who are ultimately diagnosed with epilepsy. Many of these are described as clonic-tonic-clonic seizures due to the crescendo myoclonus that ultimately culminates in a GTC seizure. Up to 50 % may have subtle lateralizing signs either in the clinical semiology or on the EEG. Absence seizures are less commonly encountered in JME and affect approximately 1/3 of patients. Yet, it is the myoclonus and generalized seizures that are the signature of this syndrome. JME may be elusive until convulsions are recognized. MRI brain is typically normal. Evidence of frontal dysfunction has been postulated as the underpinning for this genetic generalized epilepsy syndrome. The genetic component of JME is likely complex and polygenetic, and about 40 % of JME patients report a family history of epilepsy.
4. Patients with JME should receive treatment with antiseizure drugs. Even though our patient had only a solitary GTC seizure, the myoclonic seizures and JME syndrome imply recurrence if left untreated. Broad-spectrum drugs such as valproate, lamotrigine, and levetiracetam are useful ASDs. Topiramate and zonisamde may also be used in place of the enzyme-inducing ASDs, which can be sedating in the case of barbiturates, or can exacerbate generalized seizures in the case of carbamazepine, phenytoin, and gabapentin. Special considerations are required when treating women of childbearing age. While all the ASDs have a risk for birth defects, valproate has the highest teratogenic risk and should be avoided in young females due to major congenital malformations and cognitive dysfunction that is evident during early childhood development. In males, valproate is an effective choice due to its efficacy in treating the multiple seizure types associated with JME.
5. Most patients with JME do very well and are easily controlled with the correct medication. While control is usually obtained through AED treatment, breakthrough seizures may occur from noncompliance. Lifestyle changes are very important with the need to observe regular sleeping habits, avoid drugs and alcohol, and remain compliant with antiseizure medication. Treatment is usually rendered for life given the high percent that relapse will occur if ASDs are withdrawn. Still, approximately 15 % of cases are drug resistant and require additional treat-

ments which may include the vagus nerve stimulator or modified Atkins diet. Epilepsy surgery is not indicated for JME.

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