

JME: juvenile myoclonic epilepsy Thomas Geller, M.D.

Juvenile myoclonic epilepsy is one of the most common epilepsies (or epileptic syndromes) encountered in humans. An epileptic syndrome is a constellation of seizure type and signs and symptoms that have a common underlying cause, appearance, age of onset and prognosis for seizure control. This particular epileptic syndrome typically has an onset between age eight and 23 years, though occasionally will follow epileptic absence (staring, petit mal) seizures that were present at an earlier age. About 90 percent of patients with JME have had at least one generalized tonic-clonic (grand mal) seizure, and a third of JME patients will have a history of absences.

The unique feature of JME is the appearance of myoclonus (Latin for muscle jerk) that tends to happen in a daily pattern after awakening in the morning. The myoclonus is not recognized until after the onset of convulsions in over half of patients who have JME. The muscle jerks are sometimes ascribed to clumsiness in the mornings, nervousness or fatigue. The myoclonus typically involves the shoulders and arms, is not severely disabling, and is often ignored by patients or families until the adolescent has a convulsion. Myoclonus might cause a patient to spill his/her mouthwash or fling a spoon at the breakfast table, unexplainably. Absence seizures in JME are slightly different than those seen in typical childhood absence epilepsy in the pre-school child. The EEG shows slightly faster spike and wave discharges especially when the patient is exposed to certain frequencies of flashing lights.

JME has been estimated to account for 10 to 30 percent of all epilepsies and at least in some cases appears to be familial. There are a few dominantly inherited gene sites that have been identified, but the transmission of the epilepsy may be complicated by the involvement of several gene sites and a "risk load" in determining whether a patient has clinical seizures.

All of the small differences in the clinical development of this "juvenile" rather than "childhood" epilepsy make a distinct difference in the course of the disorder and response to medication. The onset seems to have something to do with a maturational effect of connections between the cortex of the brain and the deeper thalamus that is age-dependent. Unfortunately once these aberrant circuits develop, they rarely resolve themselves, and, even though it is a relatively easy-to-control epilepsy on medications, it usually requires lifelong treatment.

The majority of patients can be well controlled on a single drug, most commonly valproic acid or lamotrigine or possibly topiramate. The patient with JME is most likely to have a seizure upon awakening in the morning and is at higher risk after sleep deprivation, with fatigue, or after using alcohol. Since JME presents in adolescence, when all of these situations are more prevalent, and medication compliance is sometimes imprecise, the patient usually has to make an adjustment in his/her lifestyle. Also, since seizure control is usually simple with one-drug therapy, adolescents occasionally develop some denial that they have epilepsy at all. This can create a precarious situation, as withdrawal

seizures after termination of medicine can be severe and prolonged. The diagnosis of JME requires support from family members and recognition and acceptance that prolonged treatment is necessary. Once the lifestyle changes are initiated, JME patients have an excellent prognosis for employment, family life and personal or academic achievement.

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