Juvenile Myoclonic Epilepsy (JME)

Juvenile Myoclonic Epilepsy (JME) is one of the commonest forms of genetic generalized epilepsy. Onset is usually in adolescence. Seizures include early morning myoclonic jerks, generalised tonic clonic seizures (GTCS), and absence seizures. The EEG has generalized 3 Hz (3-6 Hz) spike/polyspike - wave complexes. A photic sensitive pattern is common. JME is usually a lifelong disorder, although seizures can be controlled in up to 90% of patients with treatment. Sodium Valproate is the drug of choice for seizure efficacy, but there are concerns for women of childbearing age (teratogenicity). Other drugs used (usually as second-line) are: levetiracetam, lamotrigine, clobazam, and topiramate. Carbamazepine is not recommended.

DEMOGRAPHICS

Prevalence: ~5% to 10% among all patients with epilepsies.
Age at Onset: 8-25 years; approximately 75% aged 12-18 years.
Genetics: Probably complex. A single gene has not been found to explain the entity.

SYMPTOMS AND SIGNS

Seizure Semiology:
- Myoclonic jerks on awakening (all patients), or at other times, are the defining feature.
- GTCS (approximately 90%), occurring sometimes after a series of myoclonic jerks.
- Absences (approximately 30%) are mild and often inconspicuous.
- Around 1/3 have all seizure types.

Timing:
- Myoclonic jerks are usually after awakening, but may occur at other times.

Seizure-Precipitating Factors:
- Sleep deprivation, fatigue, and excessive alcohol intake.
- Others include photosensitivity (1/3), mental stress, excitement, and mental and psychological arousal.

Neurological and Mental State: Normal

INVESTIGATIONS

EEG:
- EEG has generalized discharges of irregular 3 to 6 Hz spike/polyspike and wave.
Fragments of generalised spike/polyspike and slow wave are seen (these can sometimes look like focal discharges).
- Also, approximately 1/3 show photo-paroxysmal responses.
- Isolated focal discharges are also seen and can lead to a misdiagnosis.
- A sleep deprived EEG may increase yield if the awake EEG is normal, but it is important to remember sleep deprivation is a precipitant of seizures.

**Neuro Imaging:** Not usually required.

**DIFFERENTIAL DIAGNOSIS**
- Misdiagnosis occurs. Factors responsible include lack of familiarity with JME, failure to elicit a history of jerks, misinterpretation of absences or jerks as focal seizures, and high prevalence of focal EEG abnormalities.
- At onset, both Juvenile Absence Epilepsy (JAE) and JME may present with GTCS. JAE has prominent absence seizures, infrequent GTCS, and myoclonic seizures.
- Myoclonic seizures with other systemic neurocognitive involvement and neurological signs may imply metabolic aetiologies such as mitochondrial disorders or progressive myoclonic epilepsies.

**PROGNOSIS**
- All seizures are probably life-long, although patients may show improvement after the age of 40.
- Severity varies from mild myoclonic jerks to frequent and severe falls with the jerks, and GTCS if not appropriately treated.
- Seizures are generally well controlled with appropriate medication in up to 90% of patients.

**MANAGEMENT OPTIONS**
- Advice with regard to circadian distribution, lifestyle, and seizure precipitants are as important as drug treatment. Sleep deprivation is a major provocation for seizures.
- Sodium valproate is very often effective for seizure control, but for women of childbearing age, teratogenicity is a potential concern.
- Sodium valproate has been associated with significant concerns of teratogenicity (i.e. malformations, cognitive impairment, and Autistic Spectrum Disorder). This is particularly true at higher dosages. The risk of teratogenicity increases with increasing dosage. It is important clinicians and women of child bearing age are aware of this risk. Ideally, pregnancies in women with epilepsy should be planned and managed by a neurologist. Medication choices should be selected and discussed keeping in mind the safety of mother and foetus.
- Other drugs to consider (tailored to the patient) are: levetiracetam, clobazam, topiramate, and lamotrigine (N.B. Lamotrigine may aggravate myoclonic jerks).
- In women of childbearing age levetiracetam is increasingly used as the drug of first choice. Consultation with a Paediatric Neurologist may be helpful.
- Vigabatrin, tiagabine, carbamazepine, oxcarbazepine, phenytoin, pregabalin and gabapentin can exacerbate seizures in this syndrome.

**DISCUSSION WITH FAMILY**
- Safety
- Epilepsy Medical Record
- Potential of performing baseline educational assessment (through school counsellor)
- Drug Handout
- Driving

**LINK TO USEFUL RESOURCES**
Epilepsy Action (UK) have information for Parents on [Juvenile Myoclonic Epilepsy](https://pennsw.com.au/clinician-resources/epilepsy-syndromes/juvenile-myoclonic-epilepsy.html)
REFERENCES:
