**Pitfalls in Diagnosis of Epilepsy of Janz and its Implications**

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**Summary**

84 patients of juvenile myoclonic epilepsy (JME) of Janz were studied. Diagnosis was confirmed using clinical and electro-encephalographic (EEG) criterias. 58 (78%) patients of JME were referred as ‘refractory or uncontrolled seizures’. Ignoring myoclonic episodes and non-use of activation procedures in EEG were important reasons for diagnostic delay. Sodium valproate (VPA) or clonazepam are the drugs of choice while phenobarbitone (PB), carbamazepine (CZ), and phenytoin (PHT) are ineffective. Clinical spectrum of JME is slightly different in India. Family history of epilepsy or JME is not forthcoming and there is gross delay in the diagnosis. Other differences include age of presentation and mild cognitive impairment. All juvenile patients of generalized epilepsy, not responding to more commonly used CZ, PB and PHT should be strongly suspected for JME by carefully searching for myoclonus.

**Key words** : Myoclonic, Janz, Valproate.

**Introduction**

Juvenile myoclonic epilepsy of Janz (JME) is a syndrome seen in puberty and is characterized by bilateral arrhythmic, single or repetitive myoclonic jerks predominantly in arms and shoulders.1 It is diagnostically delayed epilepsy, which is often missed, wrongly labeled or frequently undiagnosed all over the world including India.2,3 Like other epilepsies, it is important to identify JME early, as it has a favorable prognosis. This study was undertaken to share our experience about epilepsy of Janz.

**Material and Methods**

The study was conducted on 84 cases suffering from JME, selected after screening 1026 cases of seizure disorder in the age range of 12-24 years. (Mean...
16.8±6.1 years). Criteria for Janz epilepsy were: i) typical sequence of absence and myoclonic seizures; followed by generalized tonic clonic seizures (GTCS), ii) diurnal variation of seizures (early morning) and exacerbation by sleep deprivation, hyperventilation, photo flickering and flashing, iii) normal cognition, physical examination and cranial imaging, iv) family history of epilepsy, v) characteristic EEG changes and vi) good response to valproate (VPA) or clonazepam therapy. Four of six criteria were mandatory for inclusion in the study.4

A thorough history (including evaluation of family) and detailed neurological examination was followed by electroencephalogram (EEG) using hyperventilation, sleep deprivation and photic stimulation as activation procedures. Radiological (cranial imaging), and biochemical (hepato-renal and metabolic) evaluation was done to rule out any pathology. These patients were managed on VPA in dose tailored to 20-40 mg/kg bodyweight. Clonazepam (0.01-0.10 mg/kg body weight) was added if they did not become seizure free even with maximum dose of VPA. Patients were followed up till they were free from seizures for a minimum period of 18 months. The duration of study was 5 years.

**Results**

Eighty four patients fulfilling the criterias of JME were in the age range of 12-18 years (mean 13.6 ± 3.6 years). No significant sex predilection was seen. Family history of seizures was present in 14 (16%), of which JME was confirmed in only 6 (43%). JME in siblings was observed in only one family. Early morning seizures and precipitation by inadequate sleep (attending late night marriages) was present in 62 (73.8%) patients. Referral diagnosis of ‘intractable or uncontrolled epilepsy’ was present in 56 (66.6%), while remaining 28 cases (33.3%) reported due to inadequate reduction in seizures. There were 37 (44%) patients on CBZ, 28 (33%) on PHT and 6 (7%) on VPA (inadequate dose) at the time of reporting. There were 4 patients (4.7%) on polytherapy of PB and PH, 4(4.7%) patients on VPA and PH and 5 (6%) on CBZ and VPA combination.

Mental status evaluation (Folstein’s scale) was within normal limits and no clinical evidence of cognitive dysfunction was present in any patient, though there were complaints of decline in scholastic performance in 9 cases. Pathological or radiographical investigations (cranial CT) revealed no focal lesions. EEG was abnormal in all cases. The characteristic EEG changes were in the form of paroxysms of bilaterally symmetrical, frontocentral accentuated polyspike and wave complexes, with relatively normal background activity. Activation procedures like photic stimulation and sleep deprivation exacerbated or precipitated the paroxysm. (Fig. 1). Nearly all (78) patients (93%) responded satisfactorily (>75% reduction in seizures) to VPA, while the rest (8%) responded on adding clonazepam, after failure to respond to maximum dose of VPA (40 mg/kg body weight). All patients were advised to sleep well and avoid working late hours at night. In 6 patients, we attempted to taper the VPA, following complaints of weight gain and alopecia (4 and 2 patients respectively) which had to be restarted following recurrence in myoclonic seizures.

**Discussion**

JME is a highly under recognized syndrome, often missed by physicians. Most of these patients are treated for any one component of the syndrome i.e. GTCS or absence, with drugs specific for that particular type of seizure, CBZ, PHT or PB, depending on physician’s choice. Unfortunately this results in progressive deterioration and increased seizure frequency. Uncontrolled seizure and trauma may result in decline in cognition, which may explain the complaints of decline in scholastic performance in 9 cases in the series. Family history of epilepsy or JME was not strong in our group of cases. More so, the gross delay in diagnosis, age of presentation and mild cognitive impairment make the clinical spectrum of JME slightly different in India. This is presumably due to: i) poor practice to classify this epilepsy before starting therapy, ii) poor recognition of nonconvulsive
Pitfalls in Diagnosis of Epilepsy of Janz

Modifications of sleep, abstinence of alcohol, reduction in intake of nicotine and caffeine have beneficial effects. VPA is the drug of choice. Many other drugs viz. primidone, PB and Clonazepam have been tried with variable responses of which clonazepam has been reported to produce beneficial effects. It is however one form of epilepsy where discontinuation should be strongly discouraged even if patients are seizure free for long and anti-epileptic medication should be advised to continue life long.

References


Accepted for publication : 7th November, 2001.