INTRODUCTION:

Juvenile myoclonic epilepsy (JME) is also known as Janz syndrome, is a common form of idiopathic generalized epilepsy. Under the proposal for revised classification of epilepsies and epileptic syndromes, in 1989, the Commission on Classification and Terminology of the International League Against Epilepsy defined JME as follows: "Impulsive petit mal appears around puberty and is characterized by seizures with bilateral, single or repetitive, arrhythmic, irregular myoclonic jerks, predominantly in the arms. Jerks may cause some patients to fall suddenly. No disturbance of consciousness is noticeable. Often, there are generalized tonic clonic seizures and, less often, infrequent absences. The seizures usually occur shortly after awakening and are often precipitated by sleep deprivation. Intercital and ictal EEG have rapid, generalized, often irregular spike waves and polyspike waves; there is no close phase correlation between EEG spikes and jerks. Frequently the patients are photosensitive. The disorder may be inherited and sex distribution is equal. Response to appropriate drugs is good". JME is thus usually considered as a relatively benign idiopathic generalized epilepsy syndrome. However, it is being increasingly recognized that JME is not a single disease entity by itself, but a broad group of various sub syndromes with differences in the genetic background, clinical characteristics, electroencephalographic features, response to treatment and prognosis. We attempted to study the epileptology of juvenile myoclonic epilepsy patients attending our center. Aims And Objectives: To study the clinical and electroencephalographic (EEG) profile of patients with juvenile myoclonic epilepsy Methods: 30 consecutive patients with juvenile myoclonic epilepsy attending our center were included. After detailed history, clinical examination all patients underwent routine electroencephalography. Results: The mean age of onset of seizure was 13.2 years with slight female preponderance (male-47%, female53%). Myoclonic jerks were present in all the patients; generalized tonic clonic seizures were present in 90% of patients and 7% of patients had absence seizures. Incidence of febrile seizures was higher (30%) in our patients. Family history of seizure was present in 13% of patients. Sleep deprivation was the commonest precipitating factor (57%) for seizure in our patients. Majority of the patients (67%) had good control of seizures with sodium valproate alone. 10% of patients had refractory juvenile myoclonic epilepsy. Routine electroencephalography was normal in 23% of cases. Conclusion: It is concluded that clinical features and routine electroencephalography data of our juvenile myoclonic epilepsy probands are comparable to reports from other parts of the world except for higher incidence of febrile seizure. Key Words: Juvenile myoclonic epilepsy, generalized tonic clonic seizure, Myoclonic jerks, absence seizures, electroencephalography.
This study was conducted to determine the clinical and EEG characteristics of patients with JME at a tertiary care hospital in Chennai.

**METHODOLOGY:**

This was a descriptive case series study, done at the department of neurology at Government Stanley Medical College Hospital, Chennai, Tamilnadu, India from February 2015 to February 2016.

Inclusion criteria:
1) Patients with clinical evidence of bilateral myoclonic jerks with or without generalized tonic - clonic seizure and/or absence seizures,
2) Patients with no evidence of neurological or intellectual deterioration,
3) Patients with normal CT/MRI brain.

Exclusion criteria:
1) Patients with Myoclonic jerks secondary to brain hypoxia, metabolic or degenerative,
2) Patients with evidence of neurological or intellectual deterioration,
3) Patients with abnormal CT/MRI brain.

All patients and their primary care givers attending epilepsy clinic were interviewed for inclusion and exclusion criteria. Patients fulfilling the inclusion criteria were included in the study after getting informed consent from the patient or their parents/guardian. Patients and their relatives were explained about the nature of the study. Detailed history including birth and perinatal history, developmental milestones, history of febrile seizures, central nervous system infection and trauma were obtained. Details of academic performance, a detailed family history and history of comorbidities were also obtained. Age of seizure onset was recorded for each type of seizures namely myoclonic seizures, absence seizures and generalized tonic-clonic seizure. Frequency of each seizure type was also noted. Details regarding myoclonic jerks like the body parts involved, whether single or multiple, and the time of the day in which jerks occurred were also noted. Similarly for generalized tonic-clonic seizure the time of the seizure, history of any preceding myoclonic jerks, any focal features in the semiology, the duration, and any postictal deficits were noted. Similarly for absence seizures the time of the seizure, semiology and duration were noted. We specifically enquired about the precipitating factors for seizures like sleep deprivation, psychological stress, alcohol use, photic stimulation and menstrual period for each patient. Details of antiepileptic drugs tried and their maximum dose, response to treatment, and side effects were noted. History of seizures in family members was also obtained.

All patients underwent one hour electroencephalography recording (both sleep and awake record). It also included activation procedures in the form of hyperventilation for 3 minutes and photic stimulation (1-50Hz for 5 seconds at a stretch with eyes open and closed). For patients who were already on treatment, the drug regime was not changed. In EEG the background activity, the frequency and duration of generalized spikes, polyspikes and slow wave discharges were noted. Asymmetry was defined as consistently unilateral epileptiform discharges, focal slowing, or a persistent unilateral predominance of the synchronous polyspike and wave discharges.

**Statistical method**

All these data were coded and entered into excel sheet and detailed analysis of the data was done by using SPSS-PC windows version 16.0. The Pearson Chi-Square test and student independent ‘t’ test were used wherever applicable and P-value less than 0.05 was taken as significant.

**RESULTS:**

Our study included 30 patients of JME, 14(46.7%) patients were males and 16(53.3%) patients were female. The mean age of study population was 22.1 years. The mean age of onset of first seizure was 13.17 years. The age of onset of first seizure ranged from 8 years to 24 years. Mean age at onset of myoclonic seizures, GTCS and absence seizures was 13.43, 14 and 8 years respectively. In this study absence seizure was present only in 2 (6.7%) patients, generalized tonic-clonic seizure in 27 (90%) patients and myoclonic jerks were noticed in all 30 (100%) patients. In this study majority of patients (26 patients, 86.7%) had both myoclonic jerks and generalized tonic-clonic seizure. 2 (6.7%) patients had only myoclonic jerks. 1 (3.3%) patient had both myoclonic jerks and absence seizure. All three types of seizures (myoclonic jerks, absence seizure, generalized tonic-clonic seizure) were present in only one (3.3%) patient. (Table – 1)

In this study first presented seizure type was absence seizure in 2 (6.7%) patients, myoclonic jerks in 10(33.3%) patients. 18 (60%) patients presented with both GTCS and myoclonic jerks in their first presentation itself. Febrile seizure was noted in 10 (33.3%) patients. Family history of seizures was present in 4 (13.3%) patients. Among the above 4 (13.3%) patients 2 were twin sisters, both of them having Juvenile myoclonic seizure.
Table -1: Seizure type distribution

<table>
<thead>
<tr>
<th>Type of seizures</th>
<th>No of patients</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myoclonic jerk</td>
<td>30</td>
<td>100%</td>
</tr>
<tr>
<td>GTCS</td>
<td>27</td>
<td>90%</td>
</tr>
<tr>
<td>Absence seizures</td>
<td>2</td>
<td>6.7%</td>
</tr>
<tr>
<td>Myoclonic jerk only</td>
<td>2</td>
<td>6.7%</td>
</tr>
<tr>
<td>Myoclonic jerk +GTCS</td>
<td>26</td>
<td>86.7%</td>
</tr>
<tr>
<td>Myoclonic jerk + Absence</td>
<td>1</td>
<td>3.3%</td>
</tr>
<tr>
<td>Myoclonic jerk +GTCS + Absence</td>
<td>1</td>
<td>3.3%</td>
</tr>
</tbody>
</table>

**PRECEPITATING FACTORS**

In majority of the patients (17 patients, 56.7%), seizure was precipitated by sleep deprivation. Fatigue precipitated seizures in 5 (16.7%) patients. Fever precipitated seizure in 1 (3.3%) patient. 13(43.3%) patients did not have any particular precipitating factor.

**RESPONSE TO MEDICAL THERAPY**

In this study 27(90%) patients had well controlled seizures with antiepileptic drugs. Out of this, 18 patients were only on sodium valproate. 7 patients needed other antiepileptics as add on with sodium valproate to control seizure. 2 patients were started on levetiracetam as initial drug since both of them were in child bearing age. 3 patients were refractory to antiepileptics inspite of trial of multiple appropriate antiepileptic.

**ELECTROENCEPHALOGRAPHY (EEG)**

In our study, 23 patients had abnormality in routine electroencephalography and 7 patients had normal records in routine electroencephalography. Out of these 23 patients, 22 patients had generalized frontal dominant 4-6Hz spike/polyspikes and slow wave discharges. One had fragmented spikes. On hyperventilation 9 patients had typical 4-6Hz spikes/polyspikes and slow wave discharges. All these 9 patients also had abnormality in routine electroencephalography even without hyperventilation. None of the patients showed abnormal response to photic stimulation.

**DISCUSSION:**

In our study the youngest patient was 8 years old and the oldest patient was 58 years of age. The mean age of study population was 22.1 years. It is comparable to other studies4,14,16. Our study showed slight female predominance (53.3%) in juvenile myoclonic epilepsy patients. Gender distribution is considered to be equal but few studies have shown female predominance16. Majority of juvenile myoclonic epilepsy patients had their first seizure between the age of 12 and 18 years4,14. In our study mean age of onset of first seizure was 13.2 years.

In our study the mean age of onset of myoclonic jerks and generalized tonic-clonic seizure were 13.5years, 14 years respectively, which is similar to previous study results4,14,15,16. However, in our study the mean age of onset of absence seizure was 8 years, which is little earlier than the onset compared to the previous studies.15,16 2 of our patients started with typical absence seizures at the age of 8 years. This is probably related to the relevant syndromic overlap between juvenile myoclonic epilepsy and childhood absence epilepsy.

In our study myoclonic jerks occurred in all patients but generalized tonic-clonic seizure occurred in 90% of patients and absence seizures in 6.7% of patients. These results are similar to the previous studies4,14.

In our study myoclonic jerks was the only type of seizure in 6.7% patients (2 patients). Majority of the patients (86.7%, n=26) had both myoclonic jerks and generalized tonic-clonic seizure. Only 1 patient (3.3%) presented with all three type (myoclonic jerks, generalized tonic-clonic seizure, absence seizure) of seizures. Both myoclonic and absence seizures were present in 1 patient (3.3%), without generalized tonic-clonic seizure. This study result is similar to the previous study15 . These results confirm that a small percentage of patients with juvenile myoclonic epilepsy can present with myoclonic jerks alone, hence high degree of suspicion is needed to identify such group of patients.

In our study the precipitating factor for juvenile myoclonic epilepsy was found in 57% patients. Sleep deprivation, either alone or in combination, was the most important precipitating factor. It was also absorbed in other studies4,15. Fatigue was reported in 16.7% (n=5) patients and fever in 3.3% (n=1) patients. Though alcohol consumption, menstrual periods, periods of concentration and stress were reported as precipitating factors in previous studies4,14,15,16 in our study none of the patients reported the same. In our study 33.3% (n=10) of patients had history of febrile seizures before the age of 5 years. In contrast to previous studies8,13, we found a high incidence of febrile seizures in our juvenile myoclonic epilepsy patients. This variation may be due to ethnic variations or ascertainment bias as isolated events early in life may be forgotten.In our study positive family history was noticed in 13.3% (n=4) patients. Incidence of family history was lower in our study as compared to previous studies. In other studies4,8,12,14 family history of seizure was noteded in 25 to 45 % of patients.
In our study 10% (n=3) patients had drug refractory juvenile myoclonic epilepsy. In the study done by Gellisse et al.11., 15.5% of patients were drug refractory. In our institution lamotrigine and topiramate were not available at the time of study period. Hence these drugs were not tried in our refractory juvenile myoclonic epilepsy patients. In our study majority (60%, n=18) of the patients had good control of seizure with sodium valproate alone, 23.3% (n=7) needed other anti epileptic drugs (clobazam in 2 patients, levetiracetam in 3 patients, phenobarbitone in 2 patients) along with sodium valproate to control seizures. 2 (6.7%) patients were started on levetiracetam as initial drug since both of them were in child bearing age and seizures were well controlled with above drugs.

Juvenile myoclonic epilepsy is an electroclinical syndrome. Though juvenile myoclonic epilepsy has the characteristic electroencephalography findings of frontocentral dominant generalized 4-6 Hz spikes/polyspikes and slow waves, various studies reported that upto 20% of patients had normal electroencephalography in routine recordings. In our study also routine electroencephalography was normal in 23.3% of our juvenile myoclonic epilepsy patients. In our study 30% had abnormal electroencephalography response to hyperventilation, but all these patients also had abnormalities in routine electroencephalography even without hyperventilation. Though most of the previous studies showed abnormal photoparoxysmal responses (nearly 10 to 20%) in their patients, in our study none of the patients had an abnormal photoparoxysmal responses. Intermittent photic stimulation, not provoking abnormalities is a significant finding in our study. However a perceptible reason could not be ascertained for the same. In our study, activation procedures did not provide any added advantage over routine awake and sleep recordings in identifying patients with juvenile myoclonic epilepsy.

CONCLUSION:

In our study majority of our juvenile myoclonic epilepsy patients had onset of seizures between 10 and 15 years of age and there was a slight female preponderance. Juvenile myoclonic epilepsy patients were clinically identified largely by a combination of myoclonic jerks and generalized tonic-clonic seizures in our study. However a small percentage of patients presented with myoclonic jerks alone and a high degree of suspicion is needed to identify this subpopulation. Significantly, one third of our study population had a history of febrile seizures and around 13% of patients had family history of epilepsy.

Majority of the patients responded to sodium valproate alone, while few required the addition of other anti epileptic drugs to sodium valproate. A good percentage of females in our study population were in reproductive age group and sodium valproate may not be a very suitable drug for this subpopulation. Few patients of this subpopulation who were tried on alternate drug had a relatively good control of seizure, which is small but significant finding of our study. Nearly 10% of our juvenile myoclonic epilepsy patients were refractory to drugs in our study. Routine electroencephalography was normal in nearly 23% of patients with juvenile myoclonic epilepsy in our study.

REFERENCES: