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JUVENILE MYOCLONIC EPILEPSY AND NEURO REHABLITATION IN CHILDREN

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Abstract:

Juvenile myoclonic epilepsy (JME) is called an idiopathic generalized epilepsy which can be identified by generalized tonic–clonic seizures (GTCS), myoclonic jerks, absence seizures and spasms which is common in majority of the cases. Although early diagnosis of Juvenile myoclonic epilepsy (JME) still a consideration due to its multiple epilepsy and also there requires a clinical profile of EEG and sleep data of the individual to support the diagnosis. There is high spike wave discharges in sleep study of EEG of the individuals under study which shows significant increase during the sleep to awakening stage or transition phase and this is to be a specific finding using sleep data in appropriate clinical setting. Since the discovery of the anticonvulsant drugs like Valproate, lamotrigine and levetiracetam are the most commonly and are established antiepileptic drugs that are effective in broad spectrum of epilepsy diagnosis and treatment. The case study which involves the retrospective analysis of epileptic patients with Juvenile myoclonic epilepsy (JME) had functional motor impairment and also went into secondary therapies such as occupational therapy, physical therapy and sensory integration to cope up to the initial dysfunction caused due to generalized tonic–clonic seizures (GTCS), myoclonic jerks, absences seizures and spasms. The routine EEGs was recorded under clinical settings with and without administration of drugs depending upon the individuals and it was found to be abnormal in 68% of cases only and the data was incorrect or misleading findings in 9%.The population under study showed significant improvement with antiepileptic drugs and secondary therapies..

Keywords **—Juvenile myoclonic epilepsy (JME), generalized tonic–clonic seizures (GTCS), occupational therapy, physical therapy, sensory integration.**

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I. INTRODUCTION

In the review Juvenile myoclonic epilepsy (JME) has the important electro clinical and possible sub syndromes evidences with genetical background which correlates with its pathophysiology and neuroimaging findings for diagnosis and treatments. The JME is one among the common types of genetic epilepsies found today. The prevalence of JME in large populations has been estimated to be between 5% to 10% of all the epileptic conditions and around 15% of the idiopathic generalized epilepsy with records varying over time and diagnosis in different settings [3, 4, 5].Albeit the condition is female predominance in many settings. Today JME is a recognized widely as electro clinical idiopathic generalized epileptic syndrome. The Onset of the epilepsy is mostly around the time of puberty in many cases. The most common ictal evidences is bilateral myoclonus which is without loss of consciousness. The JME patients show generalized tonic-clonic seizures (GTCS), and

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absence seizures. The typical circumstance of a diagnosis is first at GTCS episode, which is followed with a myoclonus or absences seizure or combination of both. The possible seizure episodes can occur after awakening from a sleep or in a relaxed stated which can be a facilitation of sleep deprivation or a sudden arousal state. The electro clinical diagnosis of JME with typical polyspike-waves or fast spike-waves on the EEG can be made the evidences GTCS. In most cases the prevalence rate of photosensitivity or photo paroxysmal EEG response in patients with JME is highly common, ranging from 6 to 60% [7] .The Hyperventilation (HV) might induce absence seizures in patients with JME, while the cognitive functioning is undisturbed in precipitation of the myoclonic seizures. Most patients tend to have a good prognosis when treated with anti-epileptic drugs, but with a high tendency to relapse after withdrawal of medications [7, 8, 9, 21]. However, there is a still considerable subgroup of JME patients whose seizures are difficult to treat and secondary therapies have helped to cope up the conditions in improving the motor and cognitive functioning of the patients. There are recent findings that suggest patients with JME have difficulty with social adjustment in different aspects of their lives, works and familiar relationship.

II. PRECIPITATION OF JME SEIZURES

The occurrence of MJ in the early morning is one of the symptoms of JME. The Myoclonic Jerks (MJ) and generalized tonic-clonic seizures (GTCS) are proven to be induced many factors such as sleep deprivation, fatigue and excessive substances abuse [15]. Sleep deprivation can be understood as falling asleep late at night and awaken early in the morning (short sleep) or getting up early. There are many seizure-provoking factors in JME among them some are stress, fatigue, fever, sleep, flashing sunlight, thinking, and excessive alcohol intake are common in adults and young teens . In JME, photosensitivity or PPR (photo paroxysmal EEG response), varies considerably [25]. However, a very small number of patients experiences seizures induced by photic stimulation in daily life. While in most cases patients with JME experiences absence seizures that are induced by hyperventilation (HV) and on the other hand myoclonic seizures are provoked during cognitive functioning [25, 26].

III. CLINICAL PRESENTATION OF JUVENILE MYOCLONIC EPILEPSY (JME)

A. Myoclonic Jerks (MJ)

Myoclonic Jerks (MJ) can occur in an individual spontaneously in the morning, after awakening from the sleep; they are characterized as short-lasting, sudden, irregular, frequently symmetric which can be isolated or appear over a clusters. These cluster when prolonged can lead to a convulsive tonic- clonic seizure. The Myoclonic jerks have muscular convulsions predominately in the upper trunk, limbs with distal and proximal movements and visible flexion and abductions of the lower and upper extremities. Patients my experiences may lose control over their extremities which may lead in dropping of things with very minimal or restricted finger movements. Whilst at many intense episode during a myoclonic –astatic seizures the patient falls down. There are several reports of authors suggests that the asymmetry nature in MJ and focal seizure features in the EEG recordings, might lead to the incorrect diagnosis of epileptic condition [26-28].

B. Generalized Tonic Clonic Seizures (GTCS)

In generalized tonic-clonic seizures (GTCS) that causes loss of consciousness and severe muscular contractions. The generalized tonic-clonic seizures (GTCS) in other words called the grandma seizures is usually due to epileptic conditions with other subsequent triggers such as diabetes, fever etc. The generalized tonic-clonic seizures (GTCS) is short lived with two major stages immediate response of loss of consciousness which occurs for about 10 to 20 second ,later sequenced by violent muscle convulsions .Whilst in most cases generalized tonic-clonic seizures (GTCS) needs anti-epileptic medications as the recurrences rate is high when stopping the medications. Frequently, in most outpatient clinic or in the emergency unit, a young

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patient is examined because of severe seizure upon awakening from the sleep.

C. Absence Seizures (AS)

Absence Seizures (AS) are generally reported in almost one-third of patients with early onset of JME [31, 32]. However, the frequency might be much higher in most cases which is 66.7% [19]. It is a generally agreed that AS associated with JME are relatively short and mild, when comparing it with the childhood absences and absences of Juvenile Absence Epilepsy. They become less significant and severe with age and are often unnoticed by the patient [33]. In a recent retrospective study, with long-term follow up of 257 patients with JME, with four major JME groups: (a) Classic JME (72%),(b) JME with Adolescent Absence (7%), (c) JME with Astatic Seizures (3%) and (d) Childhood Absence Epilepsy (CAE) evolving to JME (18%) .

D. Study method

All patients presenting to the Departments of Neurology and Paediatrics, Institute Of Child Health and Hospital for Children Tamil Nadu, India, between August 2019 and January 2020 ,with suspected epilepsy syndrome were included in the study male N= 15 , female N=20 and the following criteria suggested by Panayiotopoulos for JME was studied. The Detailed history of the onset, family medical history and neurological examination were performed. The criteria of inclusion for this study of JME: (1) the unequivocal clinical symptoms of generalized epilepsy with myoclonic jerks ; (2) the brain imaging with no structural abnormality or dysfunctioning and (3) no evidence of neurological or intellectual deficit [24]. We have excluded those with: (1) the clinical or EEG evidence of myoclonic jerks with secondary brain hypoxia, other structural brain abnormalities or metabolic disease; (2) EEG abnormalities, but no clinical evidence of any type of seizures; and (3) other generalized seizures and epileptic syndrome or possible syndrome diagnosis without firm documentation of myoclonus [24]. All the patients included in the study in according to the above mentioned criteria were subjected to a sleep EEG recording with and without induced sedatives .The sedatives were used in extreme cases to minimize movements of the subjects and the recording was done a digital EEG machine. All the patients had at least one routine EEG done before a sleep EEG was performed. However, if the patients were taking treatment, it was not stopped or changed before the study procedure. Recordings was performed using 16 channels for EEG, 2 channels was used for surface electromyogram (EMG) and 2 channels was used for electrooculography (EOG). The whole recording of EEG was reviewed, and analysed as for different stages of sleep and for spikes, poly spikes, and sharp waves as per standard practice.

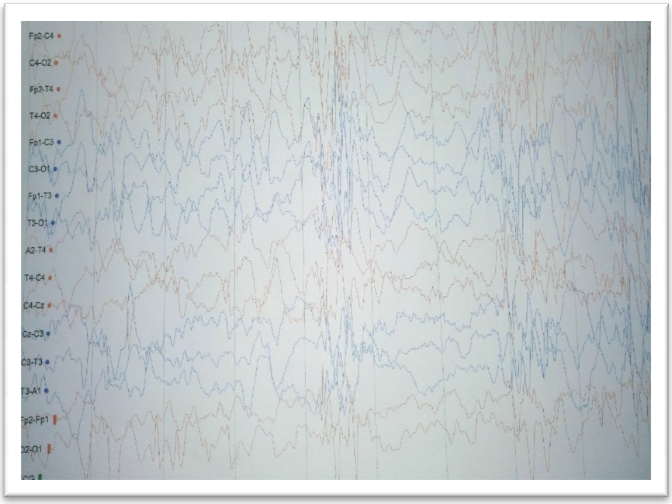

Fig. 1Interictal EEG of the patients under study shows diffuse or generalized spike-wave (SW) and poly-spike-wave (PSW) discharges at 3-6 Hz.

E. Interictal EEG

In majority of the cases localization related EEG abnormalities are found in 16-58% of patients with JME [Figure 1] [14, 27, 28, 29]. These focal abnormalities include unilateral onset with paroxysms, unilateral discharge, and frequently discharges with voltage asymmetries. In general an observation of EEG changes are predominantly seen at sleep onset and after provoked awakening [22, 23]. Photosensitivity or photo paroxysmal EEG

response in patients with JME varies and is more frequent in females and adolescents [25]

F. Occupational therapy

Occupational therapy is a secondary therapy which is an educational and practice-oriented health concerned with rehabilitation. The main role of occupational therapy is to promote an individual's participation in self-defined significant occupations, thus helps them in enabling meaningful participation in the different walk of life. In accordance with the perspective, of evaluation and useful intervention processes are achieved through the therapy. The unique contribution of occupational therapy in population with JME is in its ability to provide and enhance the possibilities of the functional limitations they experience as a result of poor seizure control. Occupations therapy for population with JME enable people to participate in various contexts of life to enhance health, well-being and quality of living.

G. Physiotherapy

Children with disorders or injuries that affect their movement or coordination are often referred to physical therapists. This imbalance is JME is due to the significant nature of the epilepsy and sever muscular contraction. Whist in most cases people with epilepsy do not need physical therapy (PT), but those with limited mobility or injury may be appropriate for PT. In these cases, physical therapists can help in enhance mobility and coordination through various approaches, including stretching, exercise, and skills development which is an appropriate intervention that various from on individual to another .The PT therapy under constant sessions help children better their movements and coordination's.

H. Sensory integration

Sensory integration is a neuro-stimulating process which can induces the sensory stimulation that contributes to behavioural and cognitive enhancement .In contrast, the sensory integrative dysfunction is a disorder in which sensory input received are not integrated or recognized in the brain and may cause varying degrees of problems in development, information processing, and behaviour. Sensory integration focuses primarily on three basic senses--tactile, vestibular, and proprioceptive, that contributes to movement and coordination of the individual. The severity of the JME can cause sensory dysfunctions in some cases. Sensory integrations has proved to be an efficient intervention in our study to help cope up with motor abnormalities

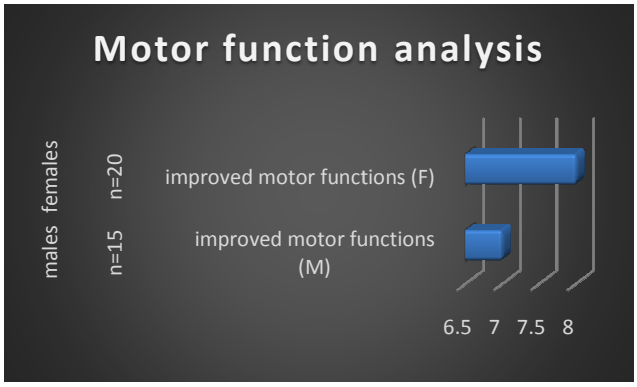

Fig. 2 Motor function analysis of the study population.

I. Results

From August 2019 and January 2020, with suspected epilepsy syndrome were included in the study male N= 15, female N=20 (fig 2), clinical diagnosis of JME was made on 35 patients. However sleep EEGs could not be conducted on 6 patients due to various reasons (usually refusal by the patient or parents) and these were excluded from the further analysis. The age ranged from 4 years to 15 years. The mean age of onset of first symptoms was 10 ± 2.513 years. The study found that successful secondary intervention therapy for patients with JME helped with improved motor functions in (Male n=7 and Female n=8) when monitored over the period of time.

IV. CONCLUSIONS

Juvenile myoclonic epilepsy JME is a common Idiopathic generalized epilepsy IGE with a significant characteristic like clinical and electroencephalograph. Generally, an EEG during sleep or an awakening EEG shall confirm the

clinical evidences. Despite the distinct EEG trait with clinical evidences, JME is often not recognized at the earliest which might result in serious consequences of late diagnosis and misleading retreatments. In most refractory cases of JME, diet such as Atkins or ketogenic might be useful which has lesser data significances. Secondary treatments like Vagus Nerve Stimulation, Deep brain stimulation, and Callosotomy are not often contemplated. Neuroimaging, uses the advanced imaging techniques, in finding structural and functional differences and changes, mainly focusing within the frontal lobes, in individuals with JME. These changes can be correlated, observed and gives the appropriate reasoning of neuropsychological deficits mainly in the frontal lobe which causes the dysfunction in patients. During the last two decades many discoveries have been made in this field of secondary assistive treatment's or neuro rehabilitation in individuals to enhance their neuronal functioning. Finding more clinical evidences on secondary treatments that have helped in motor function of the individual with the existence of specific diseases within JME.

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