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## RESEARCH ARTICLE

# PHOTOSENSITIVITY AND FAMILY HISTORY INTERRELATIONSHIP IN JUVENILE MYOCLONIC EPILEPSY (JME)

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**ABSTRACT**

Juvenile Myoclonic Epilepsy (JME) is a young onset idiopathic epilepsy syndrome characterized by Myoclonic seizures, generalized tonic clonic seizures and absence seizures. JME is an inherited disorder but the mode of inheritance is still controversial. Photosensitivity is also a precipitating factor to initiate seizures in JME. Photosensitivity is influenced by inheritance, but relation is not yet clear and established. A total 55 patients with a diagnosis of JME were evaluated to know whether triggering of seizures were influenced by photosensitivity and positive family history. JME cases were diagnosed by experienced Epileptologists on the basis of patient's medical history and on Electro Encephalo Graphy (EEG). Selection of patients and diagnosis were based on the criteria of the commission and terminology of the international League Against Epilepsy (ILAE). Patients with other neurological trauma, mental retardation and epilepsy syndrome other than JME were excluded from this study. In our study 49.09% of JME patients were photosensitive. Among these male patients were 53.12%, which was more when compared to the female i.e. 43.43%. On the other hand 43.64% of patients have positive family history. The percentage of male and female patients reporting the positive family history is almost same i.e., 43.75% in case of males and 43.47% in females. The percentage of photosensitivity and positive family history were having a close relationship, indicating that photosensitivity is linked to inheritance. So we conclude that there is a strong relationship between photosensitivity and positive family history in JME patients thus substantiating the fact that photosensitivity is inherited.



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## INTRODUCTION

Juvenile myoclonic epilepsy (JME) also mean Impulsive petit mal, characterized by seizures with bilateral, single or repetitive, arrhythmic, irregular myoclonic jerks, predominantly in the arms with Generalized tonic clonic seizures (GTCS) and less often absence seizures (Janz and Christian., 1957). The seizures usually occur shortly after awakening and are often precipitated by sleep deprivation. The term "juvenile myoclonic epilepsy" was proposed by Lund in 1975and has been adopted by the International League against Epilepsy (ILAE). The patients are photosensitive. The disorder may be inherited and sex distribution is equal (Renganathan and Delanty, 2003). Epilepsy has traditionally been referred to as a disorder or a family of disorders, rather than a disease, to imply that it is comprised of many different diseases and conditions. JME is an inherited disorder, but the exact mode of inheritance is not clear. There is a positive family history in 50% of cases or less (Renganathan and Delanty, 2003). Photosensitivity is genetically determined and occurs predominantly in females (Janz, Durner, 1997). No genes have been identified for photosensitivity. (Waltz, Stephani, 2000) Seizures triggered by photic stimulation are usually a manifestation of the idiopathic generalized epilepsies, especially JME. Photosensitivity is typically seen in 30% of

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cases, particularly in female patients (Janz., 1985), although it has been described in up to 90% of cases after prolonged photic stimulation (Appleton *et al.,* 2000). With the above facts it is obvious that there is no clear data on the inheritance of the JME, also there exists a controversy regarding the effect of photosensitivity on the JME. So the present study is planned to evaluate the inheritance pattern in JME patients and role of photosensitivity in JME.

## MATERIALS AND METHODS

The present study consist of 55 JME patients. Among 55 JME patients, 32 were males and 23 females. The age of the patients were between 8-25 years. Patient selection and diagnosis was based on the criteria of the commission and terminology of the international league against epilepsy (ILAE). JME cases were diagnosed by experienced Epileptologists on the basis of patients medical history and on Electro encephalo graphy (EEG). Patients with other neurological trauma, Mental retardation and epilepsy syndrome other than JME were excluded from this study. Questionnaire with details such as whether the individual is subjected by photosensitivity or not and family history of the patient were taken, if the family history is mentioned then whether it is paternal or maternal were recorded. Further clinical data such as type of seizure, time period of seizure were also collected. The entire study was carried out by obtaining institutional ethical clearance (IEC: MNRMC/EC/638). The data was tabulated in the excel sheet and analyzed for total number of patients experiencing the seizures by photosensitivity and those having family history, within this paternal or maternal numbers were segregated and their percentage was calculated.

## RESULTS

The present study comprises of 55 JME patients with 32 males and 23 females representing 58.2% and 41.8% respectively (Fig.1).

The present study results indicate that49.09% of JME patients were under the influence of photosensivity in experiencing the seizures. Also, the present study revealed that 53.12% of males were photosensitive. But the percentage of females was 43.43 (Table 1). The percentage of males was more when compared to females in experiencing seizures due to photosensitivity. Further, the present study on the family history of the patients indicate that 43.64% of patients have positive family history. The percentage of male and female patients reporting the positive family history is almost same i.e., 43.75% in case of males and 43.47% in females (Table 2).

The evaluation of type of inheritance in the patients who have reported positive family history indicate about 71.4% maternal inheritance and 28.6% paternal inheritance among male patients. While, it is 30% maternal and 70% paternal among female patients (Table 3 and Fig 2).

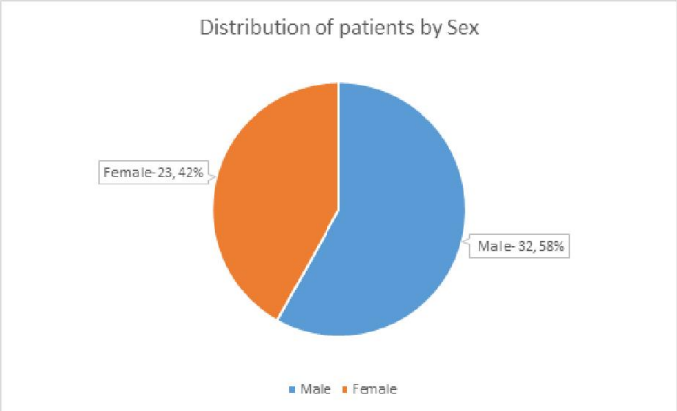


**Figure 1. Distribution of patients according to sex**

**Table 1. The percentage of JME patients affected by photosensitivity**

| | Total number | Total affected | Percentage (%) |
|---|---|---|---|
| Patients | 55 | 27 | 49.09 |
| Male | 32 | 17 | 53.12 |
| Female | 23 | 10 | 43.43 |

**Table 2. Percentage of patients with positive family history**

| Patients (n=55) | | Positive Family History | |
|---|---|---|---|
| | | Number | Percentage |
| Male | 32 | 14 | 43.7 |
| Female | 23 | 10 | 43.47 |

**Table 3. Showing number and percentage of inheritance pattern among the positive family history JME patients**

| Patients with Positive family history | Type of inheritance | | | | Type of inheritance | Total Affected | % |
|---|---|---|---|---|---|---|---|
| | | Number | % | Male JME | Maternal | 10 | 71.4 |
| | | | | | Paternal | 4 | 28.6 |
| 24 | Maternal | 13 | 54.2 | Female | Maternal | 3 | 30 |
| | Paternal | 11 | 45.8 | JME | Paternal | 7 | 70 |

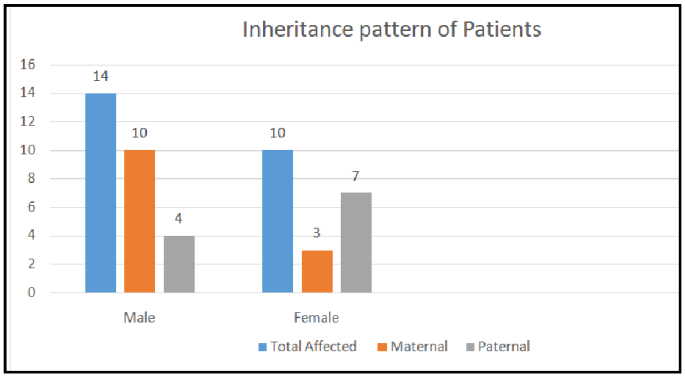


**Figure 2. Distribution of affected patients by Inheritance**

## DISCUSSION

The results of the present study indicate that 49.09% of JME patients were photosensitive. Out of these male patients under the influence of photosensitivity were 53.12% which was more when compared to the female i.e. 43.43%. In the present study prevalence of photosensitivity in juvenile myoclonic epilepsy is higher than previous reports which ranged from 8 – 45.45% i.e., by Canevini *et al.* (8%), Jain *et al.,* (9%), Julia Hofler *et al*. (14%), Janz *et al*. (30%), Panayiotopoulos *et al.* (36.8%) and Diaconu *et al*. (45.45%) and lesser than Appleton *et al*., (90%) (Janz., 1985, Appleton *et al.,* 2000, Canevini *et al.,* 1992, Jain *et al.,* 1999, Julia Hofler *et al.,* 2014, Panayiotopoulos *et al.,* 1994; Diaconu *et al.,* 2009). The difference may be attributed to geographical distribution of the samples and methodology applied. In the present study we have interviewed the patients and concluded as photosensitive if the patients are getting seizures upon variation to the light exposure. However, the findings of the present study are correlating with the reports of Yang Lu *et al.,* (2008) and Tekin Guveli *et al.* (2013). Further, Yang Lu *et al*. (2008) reported that photosensitivity was observed more in female patients i.e., 67% and less in male patients i.e., 33%. This is opposite to the findings of the present study. The explanation to this fact is difficult based on the present study but it needs further detailed evaluation to answer this. In case of family history study a total 43.64% of patientshad positive family history, among them 43.75% patients were males and 43.47% were females. This correlates with Julia Hofler *et al.,* study which reported that 41% of patients had positive family history.

Further, it can be interrelated from the present study that the percentage of patients with photosensitivity and patients with positive family history were very close indicating that all the patients with positive family history are also photosensitive which prove the point that photosensitivity is linked to inheritance. This is in line with the study by Waltz S *et al* (2000) which reports that photosensitivity is linked to inheritance. Also, the study by Durner *et al*. (2005), Greenberg *et al*. (1989), Lorenz *et al*. (2006) and Tauer *et al*. (2005) states that the human *BRD2* gene has been shown to play a susceptibility role critically in a common form of epilepsy as well as other, more subtle, neural phenomena including abnormal electroencephalographic (EEGs) patterns and photosensitivity indicating the fact that photosensitivity and inheritance are interlinked. From the present study we are reporting first time to the best of our knowledge about pattern of inheritance. Maternal path of inheritance is more among the overall patients and also among the male JME patients. But paternal inheritance is more in female JME patients. These results provide a platform for the future studies in which pattern of inheritance need to be investigated more in detail and with more sample size.

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