J Neurol Disord 2016, 4:6(Suppl)
http://dx.doi.org/10.4172/2329-6895.C1.012

Role of nicotinic acetylcholine receptor gene (839 C>T) and voltage gated sodium channel gene (563 A>T) mutations in juvenile myoclonic epilepsy (JME)

Musadiq Ahmad Bhat
Maulana Azad Medical College & Associated Hospitals, India

Juvenile myoclonic epilepsy (JME) emerges in mid-to-late childhood. The prevalence of JME in large cohorts has been estimated to be 5% to 10% of all epilepsies and around 18% of idiopathic generalized epilepsies but may be lower in some settings. Various mutations of nicotinic acetylcholine receptor gene and voltage gated sodium channel gene have been found to play a role in various types of epilepsy. In the present study, we characterized the role of 839 C>T and 563 A>T mutations of nicotinic acetylcholine receptor gene and voltage gated sodium channel gene respectively in juvenile myoclonic epilepsy (JME). The aim of the present study was to examine the role of nicotinic acetylcholine receptor gene and voltage gated sodium channel gene mutations in juvenile myoclonic epilepsy in Indian population. 3 ml of peripheral blood was collected in EDTA vials after obtaining informed consent from 50 JME patients. DNA was extracted using GeneAid DNA extraction kit and mutations of nicotinic acetylcholine receptor gene (839 C>T) and voltage gated sodium channel gene (563 A>T) in juvenile myoclonic epilepsy patients was studied by AS-PCR using primers specific for 839 C>T and 563 A>T mutations. The demographic and clinico-pathological features included the age, gender, onset of seizure, frequency of seizure, duration of seizure, postictal period, drug response, etc. For 839 C>T mutation of acetylcholine receptor gene, a statistical significant association was observed amongst the patients with clinico-pathological features like postictal period and drug response with p value of 0.0196 and 0.0377, respectively. For 563 A>T mutation of voltage gated sodium channel gene, there was a significant association of this mutation with JME (p=0.0001). However, there was no significant association of this mutation observed with any of the clinico-pathological features like onset of seizures, frequency of seizures, duration of seizures, etc. This can be concluded from the study that 839 C>T mutation of nicotinic acetylcholine receptor gene and 563 A>T mutation of voltage gated sodium channel gene may play a significant role in juvenile myoclonic epilepsy.

bhat.musadiq@gmail.com

Clinical pattern of epilepsy and their EEG finding

Riwaj Bhagat
College of Medical Sciences-Bharatpur, India

Introduction: Electroencephalogram (EEG) is a widely used diagnostic tool in epileptic patients for conforming diagnosis and optimizing drug therapy. However, there is virtually rare literature and published data on clinical pattern of epilepsy and their EEG finding in Nepal. Therefore, we attempted to study the clinical pattern of epilepsy and their EEG finding.

Methods: A descriptive retrospective study was conducted in tertiary care hospital, Chitwan, Nepal. Demographic profile, provisional diagnosis and EEG findings of epileptic patients between February 2011 and March 2014 were included and descriptive analysis was performed.

Results: The incidence of epilepsy was higher in males than in females (57.2% vs. 48.8%). The majority of patients (54.7%) referred for EEG lie in the age group of 11-30 years. More than half (61.12%) of the seizures remained unclassified. Among the classified seizure, the incidence of generalized seizure is higher than partial seizure (75.85% vs. 23.21%). Similarly, majority (55.8%) of EEG recording was found to be normal and more than one third EEG records (38.1%) were unclassified.

Conclusion: Routine twenty-minute EEGs are easy to perform and are well tolerated by patients; however, in more than half of the patients with provisional diagnosis of epilepsy, the initial EEG does not show epileptiform activity and the paroxysmal events due to its low sensitivity. Although, EEG is an essential tool for investigating epilepsy, diagnosis does not completely rely exclusively on its reading. Twenty-four hours EEG recording or Video-EEG monitoring (VEEG) may be useful tool.

forriwaj@gmail.com