

Abstract

Background: Many studies have suggested that Innate immune system and inflammatory processes can have a potential role in pathophysiology of refractory epilepsy. MEFV gene plays a major role in auto-inflammatory disorders and innate immune reactions.

Aim: The aim of this study was to determine the role of MEFV gene mutations in children with refractory epilepsy.

Methods and material: 15 refractory epilepsy patient that had attended the Neurology clinic of Bouali Children's Hospital of Ardabil were chosen according to entry criteria. The peripheral blood of the patients was collected and then the samples were screened for 12 common pathogenic variants of MEFV gene.

Results: This study included 7 girls (46.7%) and 8 boys (53.3%). The mean age of patients was 9.33 years. The mean age of patients at the onset of seizures was 1.66 years. Regarding *MEFV* gene analysis none of the patients showed mutations (0%). Based on results, it wasn't possible to determine the association was between the MEFV gene variants and, type of seizures, age and gender of the patients.

Conclusion: Our study demonstrated no association *MEFV gene mutations* and refractory epilepsy and despite the potential role of inflammatory processes and innate immune system in refractory epilepsy, according to our study this role isn't related to MEFV mutations.

Keywords: Refractory Epilepsy, MEFV Gene, seizure