Abstract

Introduction: FMF is a inflammatory and hereditary recurrent disease that is determined with self-limited recurrent fever and pain in serous, pleurtis and arthritis membranes. This disease symptoms can occure in the first 10 years of life and according to latest census more than 80% of patients, are shown this diseas symptoms in the childhood and adolescence. Gene associated with this diseas is MEFV gene that are separated from short arm of chromosome 16 (16p13). With regared to above and also high prevalence of diseas in this area, we intended to evaluate the frequency of genotypes and common alleles in these patients with designing this study.

Matherials and Methods: There is a descriptive, analytical and cross-sectional study that carried out on all persons who referred to Boo-Ali hospital in Ardabil city in 2011with familia mediterranean fever (FMF). In this study, had contacted with patients by information in patients' records and to genetic testing in terms of common mutations in MEFV gene, we refer to genetic laboratory of Imam Khomeini hospital in Ardabil city. Then obtained results from related tests with information in records and also oral interviews with patients, information were entered in to check lists wich have already been designed. Resulted data were analyzed by SPSS v16 software.

Results:in this study 300 patients were enrolled with FMF in periodic fevers clinic but during the study period, genetic analysis was performed only on 216 patients. Between all patients 167 patients (55.7%) were men and 133 patients (44.3%) were women. The age average of patients was 19.59 years and the age range was from 24 month to 18 years. cramp in 95%, fever in 93%, nausea and vomiting in 5.33%, arteritis in 4.33%, fever and chills in 3.33%, chest pain in 2.33%, each flank pain and restlessness in 1.66% of patients were seen. From 216 studied patients, 51 cases (23.61%) were with no MEFV gene mutation, 46 patients (21.29%) with heterozygous mutation, 86 patients (39.81%) with compound heterozygous genotype and 33 patients (15.27%) were with hemozygous mutation and the most common studied mutation was M694V with 23.72%. Other common mutations are V726A with 11.16%, M680I with 9.06% and E148Q with 10%. In this study were observed two heterozygous genotype, M694V-V726A with 11.11%, M694V-M694V with 8.33% and also E148Q-M694V with 5.09% are the most common genotypes between patients with gene mutation.

Conclusion: After comparing results were observed that results in this study are similar with other studies and the most common gene mutation and genotype was similar with other studies to.

Keywords: FMF, MEFV gene, Familia Mediterranean Fever