Jordan University of Science and Technology

Genotypes and Phenotypes of Arab Patients with Familial Mediterranean Fever in North Jordan

Authors:

Abstract: Objectives: Familial Mediterranean Fever (FMF) is an autosomal-recessive disorder caused by mutations in MEFV gene. Hundreds of mutations have been described in patients with FMF. The aim of this study was to analyze the common genotypes of Arab patients with FMF from Jordan and to establish genotypephenotype correlation patterns. Methods: This cross-sectional retrospective study was performed in a tertiary hospital in the north of Jordan. A total of 123 patients with FMF were recruited from the rheumatology outpatient clinic at King Abdullah University Hospital. Patients were diagnosed according to Tel-Hashomer criteria and a carrier of at least one previously identified MEFV gene mutation. Demographics and clinical manifestations were recorded. Results: Mean age at diagnosis was 17.49 years and M:F ratio was 1.05:1. The following mutations were common among our patients: R202Q (41.4%), M694V (22.1%), E148Q (10.3%), V726A (11.3%), M680I (5.3%), and M694I (3.7%). Among them 26% were homozygous, 11% were compound homozygous, 26% heterozygous, 42% were compound heterozygous, and 18% were other complex genotype including homozygosity and heterozygosity of more than one mutation. As for genotype-phenotype correlation, 65% of the patients with skin rash had R202Q mutation, while M694V mutation was associated with abdominal and chest pain. Amyloidosis correlated most with the M694I mutation (66.7%). Conclusion: The results confirm the frequency of the previously and commonly identified mutations and highlight the association of R202Q polymorphism with skin rash phenotype among adult Jordanian FMF patients. Additionally, both M694V and M694I mutations were associated with different clinical presentations