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Is MEFVGene Arg202Gln (605 G>A) A Disease-Causing Mutation?

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Abstract: Aim: Familial Mediterranean fever (FMF) is an autosomal recessive disease. Arg202Gln was reported as a frequent polymorphism, and G allele of the mutation was in linkage disequilibrium with M694V. Thus, the aim of this study was to determine the distribution of the R202Q (605G>A) mutation in exon 2 of the MEFVgene in Turkish FMF patients and controls. Materials and Methods: The study included 160 FMF and 41 FMF/amyloid patients and 121 controls. Sequencing of exon 10 and exon 5 and PCR/RFLP analysis of E148Q and R202Q mutations of exon 2 of the MEFVgene were performed for all patients according to previously described techniques. Results: We found that 5 out of 76 M694V homozygote FMF patients carry a different haplotype from the one expected. Eleven of the patients had homozygous GG allele indicating the second haplotype. None of the 121 controls was homozygous for R202Q (605G>A), but 8 controls were heterozygous for M694V mutation and 5 (4.1%) of them were in linkage disequilibrium with R202Q. Conclusions: It seems that R202Q has no effect when it is in heterozygous state; however, when combined with another disease-causing mutation, the clinical spectrum appears. Thus, R202Q might be a disease-causing mutation at least in some of the FMF patients.

Key Words: Familial Mediterranean fever (FMF), amyloidosis, MEFVgene, R202Q

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