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M694V and E148Q Mutations as Potential Molecular Markers for the Diagnosis of Familial Mediterranean Fever among Patients in the East Mediterranean Region of Turkey

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ABSTRACT The purpose of the present study was to estimate frequency of M694V and E148Q mutations in the Mediterranean Fever (MEFV) gene among different families living in the East Mediterranean region of Turkey. A total of 78 members from 19 families, who had the Familial Mediterranean Fever (FMF) disease as diagnosed in clinics, and a control group consisting of 100 members were examined in this work. The member who was clinically diagnosed with FMF gene had attracted the researchers' focus to take blood examples from the entire family members. The M694V and E148Q are point mutations located in different exons of the affected gene. It is employed PCR method with specific oligonucleotides primers pair to detect mutations in the populations. The gel electrophoresis procedure was used to visualize the presence of point mutations in FMF and control group. The M694V mutation turned to be present in 75 out of 78 members (96%) of 19 FMF-diagnosed families. Among 100 members of the control group, in 26 members 26% carried the M694V. The E148Q mutation was observed in 28 members (35.89%) of the FMF group and 8 members (8%) of the control group. To the extent of the researchers' knowledge, this is to study target E148Q mutation for FMF gene in Turkey, so this research assumed to have crucial importance in clinics to diagnose FMF gene.