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Analysis of TNFRSF1A Gene R92Q Mutation in Familial Mediterranean Fever

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ABSTRACT This study examines the frequency of TNFRSF1A gene R92Q mutation in patients with Familial Mediterranean Fever (FMF) and the role of this mutation in FMF. The study included 223 FMF patients with definite diagnosis, according to Tel-Hashomer criteria, carrying two mutations and 205 FMF patients as controls (symptomatically diagnosed) with definite diagnosis but without any of the MEFV gene mutations screened. The DNA samples of FMF patients and controls were genotyped with regard to TNFRSF1A gene R92Q mutation by PCR–RFLP method. Genotypes and allele frequencies of the TNFRSF1A gene R92Q mutation were similar in the two groups ($p=0.481$ and $p=0.48$, respectively). Because of the similarities between the symptoms of FMF and TNFRSF1A-associated periodic syndrome (TRAPS), the frequencies of the TNFRSF1A gene R92Q mutation was studied in patients with two MEFV gene mutations and also in patients without any of the twelve most common MEFV gene mutations. No significant difference was observed between the two groups. Despite sharing common symptoms, it seems that FMF is not confused with TRAPS as the TNFRSF1A gene R92Q mutation frequencies are similar between both groups.