Spectrum of mutations in the familial Mediterranean fever gene (MEFV) in Turkish patients of the Central Anatolia region: a comparison of two mutation detection system

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ABSTRACT. The purpose of this study was to determine the spectrum of the most common mutations in the familial Mediterranean fever gene (MEFV) in Turkish patients from the Central Anatolia region, by using two different methods for detecting FMF-associated mutations with different screening panels, and compare our results with other diagnostic molecular genetics centers. A total of 1579 patients were analyzed. Genomic DNA from 304 patients was tested for 6 common mutations located in exon 2 (E148Q), and exon 10 (M680I, M694V, M694I, V726A, R761H) by real-time PCR while 1275 patients were tested for 17 mutations located in exon 2 (E148Q), and exon 10 [M680I (G/C), M680I (G/A), I692del, M694V, M694I, K695R, V726A, S675N, G678E, M680L, T681I, M694L, K695M, R717S, I720M, V722M] by pyrosequencing. The most frequent mutation was M694V, followed by M680I, E148Q, and V726A. Ten mutations in the panel were not detected in any patients. Finally, we compared our results with those of other centers in Turkey to contribute to the identified spectrum of
Turkish *MEFV* mutations and we discuss which *MEFV* mutations are informative for evaluating an FMF patient.

**Key words:** *MEFV* gene; FMF; Mutation; Molecular diagnosis