

Abstract

MEFV Gene Mutations in Iranian Patients with Familial Mediterranean Fever (FMF)

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Objectives: Familial Mediterranean fever (FMF) is an autosomal recessive disease, which is characterized by recurrent short episodes of inflammation in serous membranes. It is most prevalent in Western Mediterranean population. MEFV is the only gene currently known to be associated with this disease. Previous studies revealed that 6% of Iranian Jewish residents in Israel were carriers of MEFV mutation. The present study was undertaken to analyze the three known mutations of this gene in Iranian patients.

METHODS: After signing informed consent, 30 patients meeting full inclusion criteria and 30 healthy controls enrolled in the study. To detect M694V, M680I and V726I mutations, genomic DNA, extracted from peripheral blood lymphocytes, was examined using amplification refractory mutation system (ARMS-PCR) and common, normal and mutant specific primers.

RESULTS: Fever and intermittent episodic abdominal pain were the most common manifestations among the cases. Of the 30 cases with full inclusion criteria, 9(30%) were positive for M694V mutation (2 homozygote, 6 simple heterozygote, and one compound heterozygote), 4 (13.3%) for M680I mutation (one homozygote, 2 simple heterozygote, and one compound heterozygote) and one (3.3%) for V726I mutation. All of the controls were negative for the three mutations.

CONCLUSION: As in other ethnic groups, M694V mutation is the most common MEFV mutation in Iranian FMF patients but it exists in lower frequency. Consequently we recommend full mutation analysis of MEFV gene in all of our samples especially in those we could not find any of the three mutations mentioned above.