GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Frequency of alterations in the *MEFV* gene and clinical signs in familial Mediterranean fever in Central Anatolia, Turkey

G.G. Ceylan[1], C. Ceylan[2] and E. Ozturk[1]

[1]Department of Medical Genetics,
Ankara Ataturk Education and Training Hospital, Ankara, Turkey
[2]Department of Urology,
Turkiye Yuksek Ihtisas Education and Training Hospital, Ankara, Turkey

Corresponding author: G.G. Ceylan
E-mail: nil_cey@yahoo.com

Genet. Mol. Res. 11 (2): 1185-1194 (2012)
Received September 15, 2011
Accepted February 24, 2012
Published May 7, 2012
DOI http://dx.doi.org/10.4238/2012.May.7.4

ABSTRACT. Familial Mediterranean fever is a recessive autoinflammatory disease that is frequent in Armenians, Jews, Arabs, and Turks. The *MEFV* gene is responsible for this disease. We looked for *MEFV* gene variations (polymorphism and mutations) in a population that resides in Central Anatolia, Turkey. DNA was extracted from peripheral blood leukocytes of 802 familial Mediterranean fever patients. The DNA sequence data were examined for approximately 150 different mutations and polymorphisms, including single nucleotide polymorphisms in different exons of the *MEFV* gene. The male:female ratio of these patients was 1.44:1. Mutations were detected in 48.1% of the patients; 7.5% were homozygous, 11.1% were compound heterozygous and 31.5% had only one identifiable mutant allele. No mutations were detected in 51.9% of the patients. The main clinical characteristics of the patients were: abdominal pain in 20.6%, arthritis in 22.9%

and amyloidosis in 4.6%. Sixty-six percent of patients had a family history of familial Mediterranean fever; 19.4% of the patients were found to have parental consanguinity. We conclude that the genetics of familial Mediterranean fever is more complex than has previously been reported; heterozygous patients presenting a severe phenotype should be further analyzed for less common secondary MEFV mutations, using gene sequencing.

Key words: *MEFV* gene; DNA sequencing; Genetics; Mutation; Familial Mediterranean fever

