GENETIC AND RENAL CLINICAL CHARACTERIZATION OF FAMILIAL MEDITERRANEAN FEVER IN ARMENIAN POPULATION

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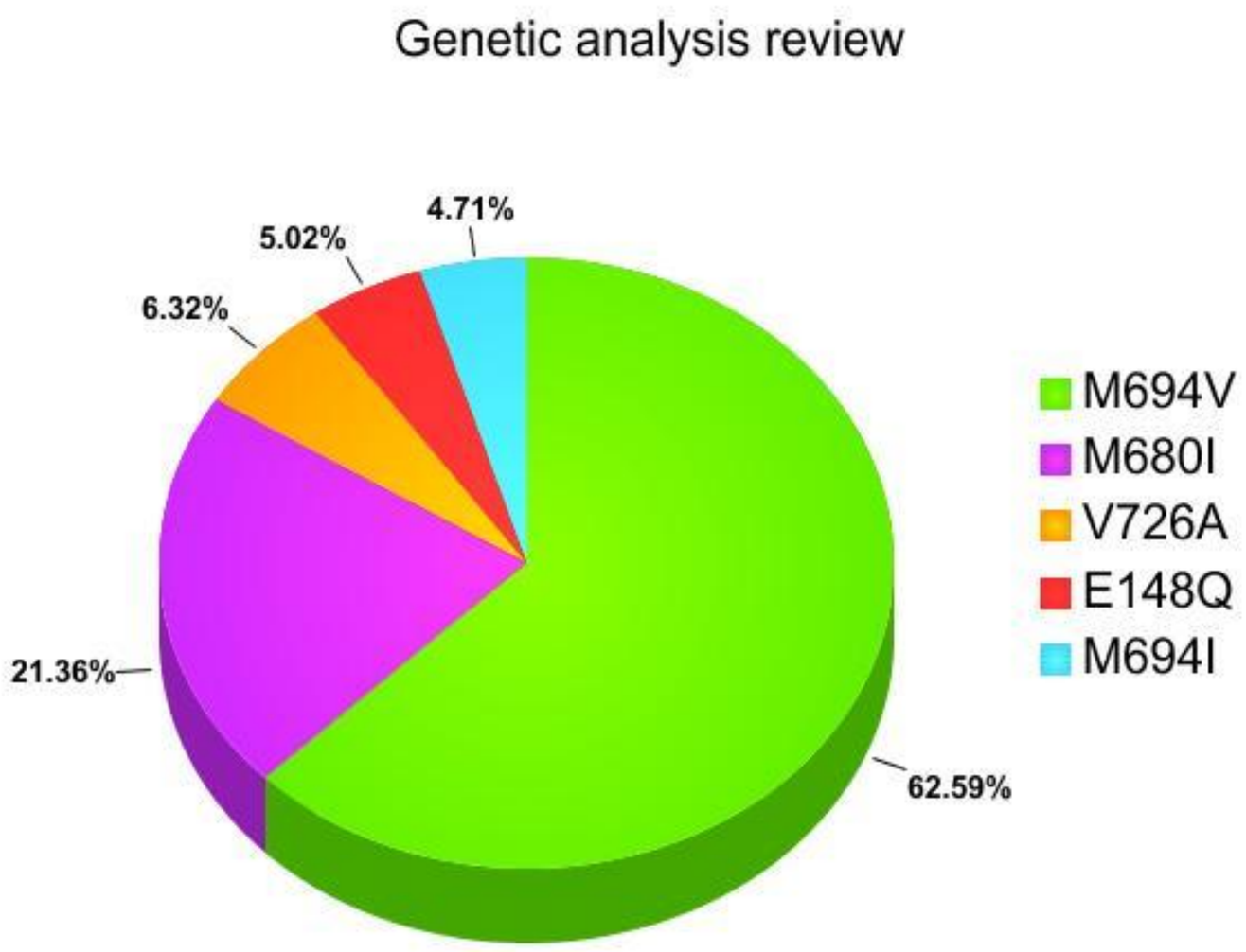
OBJECTIVES

Familial Mediterranean Fever (FMF), also known as Armenian disease, is an autosomal recessive disease affecting mainly Mediterranean populations as Armenians, Jews, Arabs, Turks. Amyloidosis, causing renal failure, is one of the most severe complications of the disease. The MEFV gene responsible for the disease has been identified on 16p13.3. It is composed of 10 exons and spans approximately 14 Kb of genomic DNA. Hitherto more than 35 mutations of this gene have been identified. From which the most frequent are M694V, M694I, M680I, E148Q and V726A. The aim of this study was to investigate the role of genetic factors on the phenotype and on the development of renal amyloidosis in a large group of patients. We conducted a phenotype/genotype correlation study in a population of Armenians with FMF.

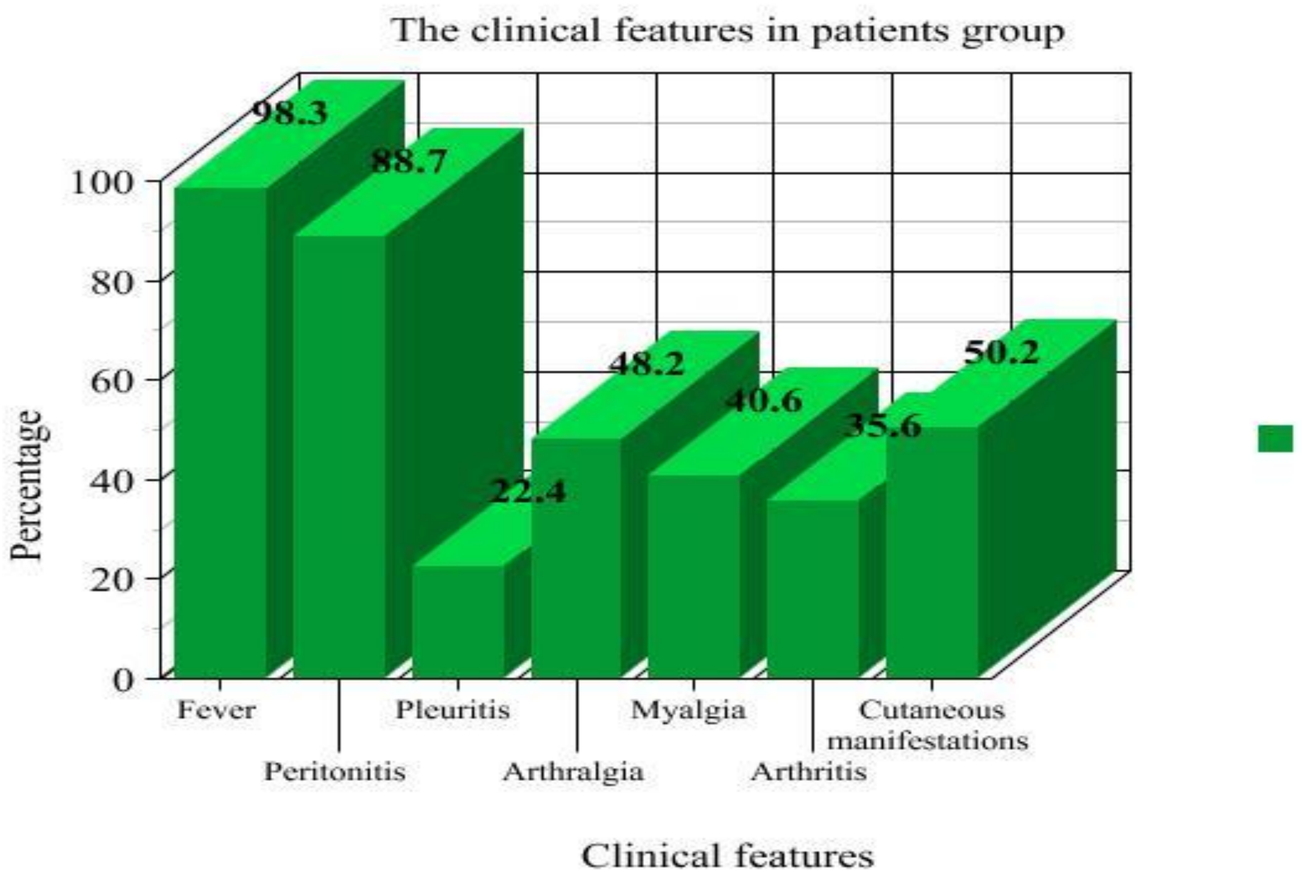
METHODS

Five hundred patients clinically diagnosed with Familial Mediterranean Fever underwent molecular genetic studies using polymerase chain reaction (PCR) methods to detect the presence of the mutations (M694V, M680I, V726A, E148Q , M694I). We compared the clinical characteristics of the inflammatory attacks in patients, while controlling for the type of mutation, correlated the presence of each mutation with clinical manifestations, disease severity, and occurrence of renal amyloidosis. The cohort was composed of 500 patients with age range of 18-70yr, with a male:female ratio of 1.5:1.



RESULTS

The M694V mutation was the most frequent mutation in this cohort study (62.59%). In patient group with homozygous M694V mutations renal amyloidosis was present in 48.5%, in heterozygous patients group 10.8%. The M680I mutation was present in 21.36%, V726A 6.32%, E148Q 5.02% and M694I 4.71%.
The clinical features in patients group included fever (in 98.3%), peritonitis (88.7%), pleuritis (22.4%), arthralgia (48,2%), myalgia (40.6%), arthritis (35.6%), also cutaneous manifestations in 50.2% including erysipelas-like erythema and oral ulcers. In patients group with E148Q gene mutations clinical features were mildly presented.



CONCLUSIONS

As a result in our study, M694V was found to be the most prevalent mutation in Armenian population and was associated with intensity of peritonitis, fever and cutaneous manifestations.

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