

Epidemiology of Familial Mediterranean fever mutations in Georgia

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Introduction

Familial Mediterranean fever (FMF) is an inherited disorder characterized by recurrent episodes of fever accompanied by sterile peritonitis, arthritis, and pleuritis. This disease is caused by mutations in MEFV gene and it's inherited in autosomal recessive manner.

Aim

Our aim was to gather epidemiological information in Georgia, try to find relativity between specific mutations and the severity of disease and also to compare this information with the relative studies of other countries.

Methods

We have analyzed results of genetic testing, which was made by 2nd and 10th exomes sequencing in MEFV gene, after which M694V, M680I, E148Q, V726A, R761H, E251K mutations were checked manually. Volunteers also filled the form that included information about: their age, dosage of Colchicine per day, amount of attacks per year, their age when first clinical symptoms appeared.

Results

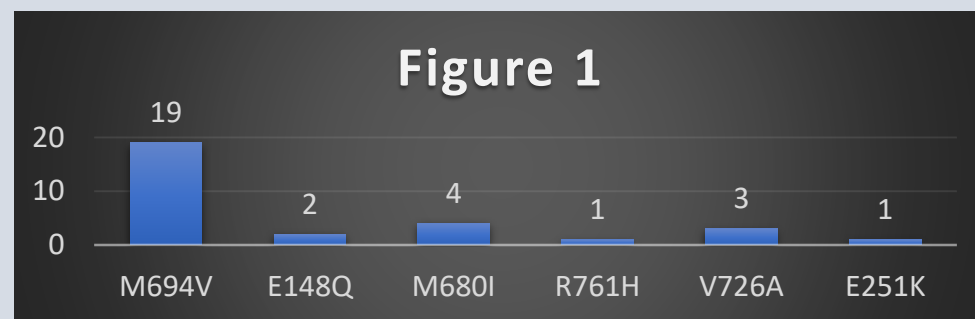


Figure 1 shows distribution of MEFV alleles among 15 patients in Georgia

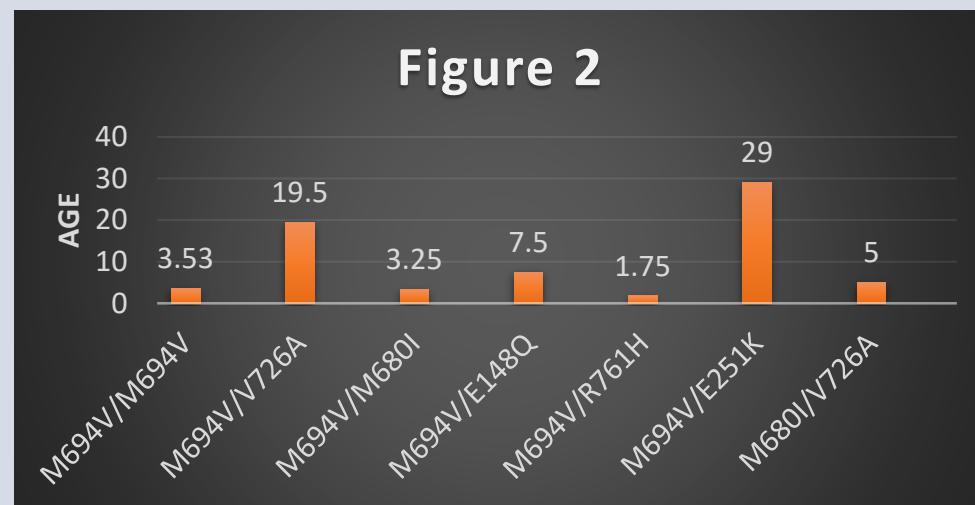


Figure 2 shows average age of clinical manifestation dependent on specific pairs of mutations.

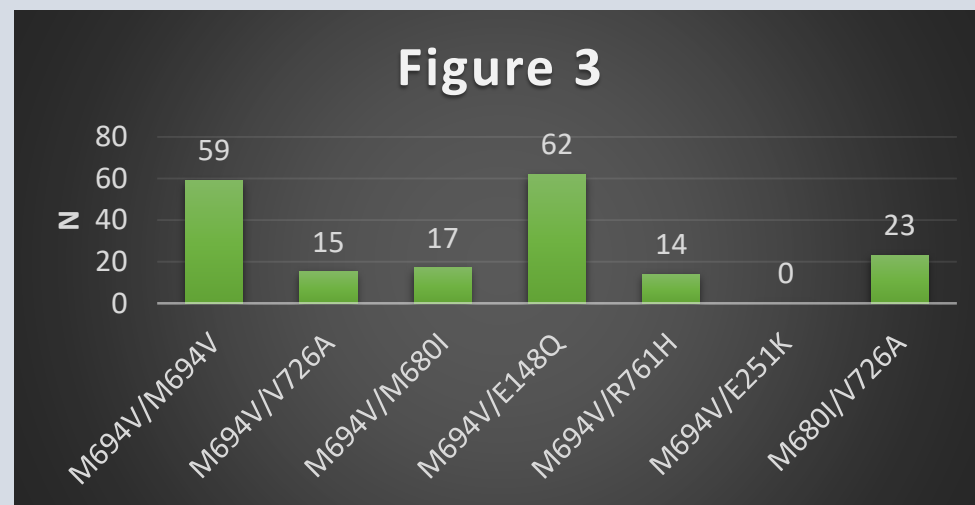


Figure 3 shows controllability of disease via Colchicine dependent on MEFV alleles.

$$N = \frac{\text{Dosage} * \text{appearances per year}}{\text{age}}$$

Smaller is the N, better life quality has patient with the help of Colchicine.

Conclusion

Clinical manifestation and life quality varies between certain allele pairs of MEFV gene, so regarding this fact, case management should be individual. Also studies showed that M694V is prevalent mutation in Georgia, that is quite unique.