



Evaluation of Clinical, Genetic and Treatment-Related Characteristics in FMF Patients by Gender Distribution

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Turk J Int Med 2021;3(Supplement 1):S29-S30

DOI: [10.46310/tjim.876499](https://doi.org/10.46310/tjim.876499)

Keywords: FMF, Gender, MEFV

Individuals with familial Mediterranean fever (FMF) may be exposed to stress due to gender-related differences and consequently the frequency of attacks may be different. For example, FMF attacks can be triggered in women during menstrual periods.^{1,2} The aim of this study is to investigate the differences between males and females in clinical findings, hereditary characteristics, treatment responses and pathogen Mediterranean fever (MEFV) gene phenotype frequencies in FMF patients.

The charts of 213 patients who were followed up in the rheumatology outpatient clinic with a diagnosis of FMF were retrospectively reviewed. The data of 105 patients (70 females, 35 males) whose charts were available for all research data were evaluated. While evaluating the clinical findings; The age of attack onset, attack character (typical, atypical), dominant attack location (peritoneum, pleura, synovia, isolated fever), presence of recurrent fever, appendectomy

history, family history (first degree, second degree) were evaluated. While evaluating the treatment response, the response (complete, partial, unresponsive) to colchicine treatment was questioned. The phenotype frequencies of the pathogen variations (M694V, V726A, M680I, E148Q) in the MEFV gene were determined. Findings were compared between groups.

The median age (minimum-maximum) in women and men was 37.5 (19-62) and 30.0 (19-59) years, respectively (p=0.148). Demographic characteristics, clinical findings, treatment responses of the participants are summarized in Table 1. The frequency of individuals with typical attack character was 71.4% in women and 82.9% in men. The frequency of the predominant attack localization with peritoneum was 90% in women and 88.6% in men. The frequency of recurrent fever in women and men was 67.1% and 65.7%, the frequency of appendectomy was 34.3% and 42.9%, and the presence of a family history was



Received: February 08, 2021; Accepted: March 4, 2021; Published Online: March 6, 2021

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Table 1. Evaluation of clinical, hereditary and treatment-related characteristics of individuals with Familial Mediterranean fever disease by gender distribution

Variables	Female (n=70)	Male (n=35)	p value
Age (years)	37.5 (19-62)	30.0 (19-59)	0.148
Attack onset age (years)	15.5 (1-50)	16.0 (4-43)	0.965
Clinical, hereditary and treatment-related findings, n (%)			
Attack character			0.201
Typical	50 (71.4)	29 (82.9)	
Atypical	20 (28.6)	6 (17.1)	
Dominant attack localization			0.557
Peritoneum	63 (90.0)	31 (88.6)	
Pleura	1 (1.4)	0 (0)	
Joint	5 (7.1)	2 (5.7)	
Isolated fever	1 (1.2)	2 (5.7)	
Relapsing fever	47 (67.1)	23 (65.7)	0.884
Appendectomy	24 (34.3)	15 (42.9)	0.392
Family history	53 (75.7)	27 (76.1)	0.175
First degree	40 (57.1)	25 (71.4)	
Second degree	13 (18.6)	2 (5.7)	
Colchicine response			0.427
Good	57 (81.4)	25 (71.4)	
Limited	8 (11.4)	5 (14.3)	
No	5 (7.2)	5 (14.3)	

Table 2. Comparison of phenotypic frequencies of MEFV gene mutations according to gender distribution of individuals with Familial Mediterranean fever disease

Phenotype frequency (n, %)	Female (n=70)	Male (n=35)	p value	OR (CI)
M694V	42 (62.7)	31 (88.6)	0.006	4.613 (1.456-14.613)
V726A	15 (22.4)	5 (14.3)	0.328	0.578 (0.191-1.749)
M680I	14 (20.9)	8 (22.9)	0.819	1.122 (0.419-3.002)
E148Q	10 (14.9)	5 (14.3)	0.931	0.950 (0.298-3.033)

MEFV: Mediterranean fever, OR: Odds ratio, CI: confidence interval.

75.7% and 76.1%, respectively. The proportion of those with colchicine response was 92.8% and 85.7% in women and men, respectively. Clinical findings and colchicine response were not different between genders. The phenotype frequency of pathogen MEFV gene mutations are summarized in Table 2. The phenotype frequencies of pathogen MEFV gene mutations were 62.7% and 88.6% for M694V ($p=0.006$), 22.4% and 14.3% ($p=0.328$) for V726A, 20.9% and 22.9 for M680I ($p=0.819$) and 14.9% and 14.3% ($p=0.931$) for E148Q in women and men, respectively.

Clinical findings and treatment responses are not different in individuals with FMF disease. The frequency of the M694V mutation, which has high penetration and is associated with important complications such as amyloidosis, is higher in men. There is a need for studies to evaluate FMF activity according to gender distribution.

Conflict of Interests

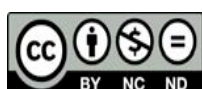
Authors declare that there are none.

Acknowledgment

This study has been presented in 17th Uludag Internal Medicine National Winter Congress, 6th Bursa Family Medicine Association National Congress, 11th Uludag Internal Medicine Nursing Congress, 5–7 March 2021, Bursa, Turkey.

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