

POSTER PRESENTATION

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Featuring the phenotype of the FMF prototype

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Background

The presentation of FMF is extremely variable, ranging from a quiescent to severe disabling disease. The M694V mutation is one of approximately 300 published genetic variations of MEFV and is thought to be associated with a typical clinical picture of the disease, but studies featuring the phenotype of homozygous M694V phenotype are meager.

Objectives

To describe the clinical trait of M694V homozygous FMF as compared to the phenotype of FMF with mixed MEFV genotypes.

Patients and methods

Fifty seven FMF patients homozygous for the M694V genotype were compared to 56 patients carrying other mutations. A questionnaire, including items related to demographic and clinical features was completed for each patient based on interview, physical examination and file notes.

Results

Compared with the control group, more patients, homozygous for the M694 mutation, suffered from a severe disease (p=0.001), had higher frequency of attacks before and during colchicine treatment (p=0.0001 and 0.0007, respectively), had more related diseases (p=0.0373) and needed higher dose of colchicine to control their disease (p=0.0001). Most other features tested (Table 1) appeared to be more pronounced in M694V homozygous patients (either with or without statistical significance).

Conclusion

The phenotype of FMF, as manifested in M694V homozygous patients, is the gold standard, to which other FMF presentations should be compared.

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Table 1

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Parameter	694 Homozygous (N=57)	Other mutations (N=56)	Р
Average length of attack (days)	2.66±1.5	3.03±1.2	0.073
Abdominal attacks	50 (87.7%)	48 (85.7%)	0.788
Arthritis attacks	52 (91.3%)	28 (50%)	<0.0001
Pleuritis attacks	36 (46.2%)	18 (38.2%)	0.0013
Exertional leg-pain	47 (82.5%)	36 (64.3%)	0.034
ELE attacks	10 (17.5%)	3 (5.4%)	0.073
Attacks of fever alone	20 (35.1%)	12 (21.4%)	0.143
Average colchicine dose (mg/day)	1.9±0.48	1.48±0.54	0.0001
IV colchicine treatment	5 (8.8%)	0 (0%)	0.057
Proteinuria or amyloidosis	6 (10.5%)	1 (1.8%)	0.113
Anemia of chronic disease	14/53 (26.4%)	7/52 (13.5%)	0.142
Elevated acute phase reactants	10/18 (55.6%)	4/16 (25%)	0.092
Chronic renal failure	6 (10.5%)	0 (0%)	0.027
Chronic arthritis	11 (19.3%)	2 (3.6%)	0.015
Work days lost each month	4.4±7.2	2.6±4.6	0.718
Harm to quality of life	(1-10) 5.6±3.3	4.1±3	0.013
Number of attacks per year w colchicine	7.2±7.8	3.5±5.5	0.0007
Number of attacks per year w/o colchicine	23.6±9.3	15.6±11.7	0.0001
Crohn's disease	4 (7%)	2 (3.6%)	0.679
Ankylosing Spondylitis	3 (5.3%)	1 (1.8%)	0.619
Behcet's Disease	7 (12.3%)	1 (1.8%)	0.061
Henoch Schonlein Purpura	1 (1.8%)	0	1
All FMF associated diseases	17 (29.8%)	7 (12.5%)	0.0373

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