

Supplementary Table S1. The *MEFV* gene mutations in patients with familial Mediterranean fever.

Patient ID	Exon	Allelic expression in exon 10	<i>MEFV</i> gene mutation	Classification [†]
P1	10	Biallelic	M694V/M694V	Pathogenic
P2	10	Biallelic	M694V/M694V	Pathogenic
P3	10	Biallelic	M694V/M694V	Pathogenic
P4	10	Biallelic	M694V/M694V	Pathogenic
P5	10	Biallelic	M694V/V726A	Pathogenic
P6	10	Biallelic	M694V/V726A	Pathogenic
P7	10	Biallelic	M694V/M680I	Pathogenic
P8	10	Biallelic	M680I/M680I	Pathogenic
P9	10	Monoallelic	M694V/-	Pathogenic
P10	10	Monoallelic	M694V/-	Pathogenic
P11	10	Monoallelic	M694V/-	Pathogenic
P12	10	Monoallelic	M680I/-	Pathogenic
P13	10	Monoallelic	M680I/-	Pathogenic
P14	10, 2	Monoallelic	M694V/E148Q	Pathogenic
P15	10, 2	Monoallelic	V726A/E148Q	Pathogenic
P16	2	-	E148Q/-	Uncertain significance

MEFV: *MEditerranean FeVer*.; [†]Van Gijn ME, et al. New workflow for classification of genetic variants' pathogenicity applied to hereditary recurrent fevers by the international study group for systemic autoinflammatory diseases (INSAID). *J Med Genet*. 2018;55(8):530-537.

Supplementary Table S2. Comparison of clinical and laboratory data according to the *MEFV* gene mutations[†].

	Biallelic 'exon 10' mutation (n=8)	Monoallelic 'exon 10' mutation with or without <i>E148Q</i> (n=7)	<i>p-value</i>
Disease onset in years, mean $\pm$ SD	3.3 $\pm$ 2.0	3.4 $\pm$ 2.7	0.896
Total ISSF score, mean $\pm$ SD	3.9 $\pm$ 1.9	2.3 $\pm$ 1.3	0.075
Clinical findings of familial Mediterranean fever, n (%)			
Fever	8 (100)	7 (100)	1.000
Abdominal pain	7 (87.5)	6 (85.7)	1.000
Chest pain	3 (37.5)	1 (14.3)	0.569
Attack visit, median (min-max)			
CRP, mg/L	45.1 (9.1-94.8)	18.6 (11.0-99.6)	0.817
SAA, mg/dL	54.9 (10.3-127.0)	18.6 (2.9-63.5)	0.247
IL-1 β , pg/mL	2.3 (1.4-10.2)	2.4 (1.6-6.7)	1.000
IL-18, pg/mL	44.9 (21.8-165.3)	42.0 (34.3-99.9)	0.908
GSDMD- $\Delta^{\ddagger}$	7.0 (2.1-61.7)	8.2 (5.3-54.6)	0.562
Attack-free visit, median (min-max)			
CRP, mg/L	2.9 (0.7-5.2)	1.0 (0.6-6.6)	0.816
SAA, mg/dL	0.7 (0.3-7.9)	0.3 (0.2-4.2)	0.296
IL-1 β , pg/mL	2.5 (1.7-10.0)	2.5 (1.6-6.3)	0.817
IL-18, pg/mL	44.8 (29.5-158.9)	35.8 (29.5-99.9)	0.643
GSDMD- $\Delta^{\ddagger}$	0.5 (0-6.5)	0.8 (0.1-5.1)	0.726

MEFV: MEiterranean FeVer, SD: standard deviation, ISSF: International Severity Scoring System for Familial Mediterranean Fever, CRP: C-reactive protein, SAA: serum amyloid-A, GSDMD: gasdermin-D, GSDMD- Δ : gasdermin-D gene expression (‡ mean $2^{-(\Delta\Delta Ct)}$).; ‡ Normal ranges for serum acute phase reactants and interleukins: CRP<5 mg/L; amyloid-A<0.5 mg/dL; IL-1 β <12 pg/mL; IL-18<120 pg/mL.