

Supplementary Table S1. Symptoms of 54 adult patients with CAPS according to the majority subtypes.

Symptoms, n (%)	FCAS (n=14)		MWS (n=30)		CINCA (n=7)	
	Childhood	Last year	Childhood	Last year	Childhood	Last year
Fever	10 (71)	2 (14)	23 (79)	7 (23)	6 (86)	3 (43)
Abdominal pain	0 (0)	0 (0)	7 (23)	3 (10)	1 (14)	2 (29)
Nausea and vomiting	0 (0)	0 (0)	8 (27)	1 (3)	0 (0)	7 (100)
Diarrhoea	0 (0)	0 (0)	4 (13)	5 (17)	3 (43)	3 (43)
Chest pain	0 (0)	0 (0)	4 (13)	2 (7)	1 (14)	1 (14)
Painful lymph nodes	1 (7)	1 (7)	8 (27)	0 (0)	4 (57)	3 (43)
Urticaria	13 (93)	5 (36)	29 (97)	3 (10)	5 (71)	1 (14)
Mouth ulcers	7 (50)	0 (0)	14 (48)	8 (27)	5 (71)	5 (71)
Myalgia	12 (86)	8 (58)	17 (57)	8 (27)	6 (86)	5 (71)
Arthralgia	14 (100)	5 (38)	26 (87)	15 (50)	6 (86)	5 (71)
Arthritis	10 (71)	2 (14)	22 (73)	7 (23)	6 (86)	5 (71)
Headache	5 (36)	3 (31)	17 (57)	10 (33)	5 (71)	4 (57)
Papillary oedema	0 (0)	0 (0)	0 (0)	0 (0)	3 (43)	1 (14)
Conjunctivitis	5 (36)	3 (21)	24 (80)	8 (28)	4 (57)	3 (43)
Episcleritis	0 (0)	0 (0)	1 (3)	2 (7)	0 (0)	0 (0)
Keratitis	0 (0)	0 (0)	0 (0)	2 (7)	0 (0)	0 (0)
Uveitis	0 (0)	0 (0)	1 (3)	0 (0)	1 (14)	0 (0)
Meningitis	0 (0)	0 (0)	1 (3)	1 (3)	4 (57)	1 (14)
Intellectual deficiency	0 (0)	0 (0)	2 (7)	3 (10)	4 (57)	4 (57)
Hearing loss	0 (0)	0 (0)	20 (67)	24 (80)	4 (57)	7 (100)
Amylosis	0 (0)	0 (0)	2 (7)	4 (13)	1 (14)	2 (71)
Growth retardation	3 (21)	4 (29)	7 (23)	4 (13)	3 (43)	2 (29)
Hypogonadism	1 (20)	2 (40)	2 (13)	4 (25)	2 (100)	2 (100)

Supplementary Table S2. Symptoms of 54 adults' patients with CAPS according to the majority variants.

Symptoms, n (%)	A439V (n=17)		R260W (n=16)	
	In childhood	Last year	In childhood	Last year
Fever	11 (65)	3 (18)	15 (100)	5 (31)
Abdominal pain	1 (6)	1 (6)	7 (44)	3 (19)
Nausea and vomiting	1 (6)	0 (0)	3 (19)	0 (0)
Diarrhoea	0 (0)	0 (0)	3 (19)	3 (19)
Chest pain	0 (0)	1 (6)	4 (25)	0 (0)
Painful lymph nodes	2 (12)	1 (6)	4 (25)	0 (0)
Urticaria	16 (94)	5 (29)	16 (100)	2 (13)
Mouth ulcers	7 (41)	1 (6)	9 (56)	5 (31)
Myalgia	16 (94)	12 (71)	9 (56)	5 (31)
Arthralgia	17 (100)	8 (47)	13 (81)	10 (63)
Arthritis	14 (82)	3 (18)	10 (63)	4 (25)
Headache	7 (41)	5 (29)	9 (56)	6 (38)
Papillary oedema	0 (0)	0 (0)	0 (0)	0 (0)
Conjunctivitis	8 (47)	3 (18)	13 (81)	4 (27)
Episcleritis	0 (0)	0 (0)	1 (7)	1 (7)
Keratitis	0 (0)	0 (0)	0 (0)	0 (0)
Uveitis	1 (6)	0 (0)	0 (0)	0 (0)
Meningitis	0 (0)	0 (0)	0 (0)	0 (0)
Intellectual deficiency	0 (0)	0 (0)	2 (13)	3 (19)
Hearing loss	3 (18)	4 (23)	10 (63)	13 (81)
Amylosis	0 (0)	0 (0)	0 (0)	1 (6)
Growth retardation	5 (29)	4 (23)	1 (6)	0 (0)
Hypogonadism	2 (40)	2 (40)	1 (11)	3 (33)

Supplementary Table S3. Flare up characteristics of all patients, main subtypes and main variants.

	All patients (n=54)	FCAS (n=14)	MWS (n=30)	CINCA (n=7)	A439V (n=17)	R260W (n=16)
Duration flare-up, n (%)						
1 day	15 (28)	5 (36)	8 (28)	1 (14)	5 (29)	4 (25)
1-3 days	17 (32)	6 (43)	9 (31)	2 (29)	8 (47)	5 (31)
> 3 days	10 (19)	2 (14)	6 (21)	2 (29)	3 (18)	4 (25)
Absence	11 (21)	1 (7)	6 (21)	2 (29)	1 (6)	3 (19)
Number per year, n (%)						
<12	23 (43)	7 (50)	12 (41)	3 (43)	10 (59)	4 (27)
12-24	8 (15)	3 (21)	4 (14)	1 (14)	4 (24)	3 (20)
>24	11 (21)	3 (21)	7 (24)	1 (14)	2 (12)	5 (31)
Absence	11 (21)	1 (7)	6 (21)	2 (29)	1 (6)	3 (20)
Duration fever, n (%)						
<12h	19 (35)	7 (50)	8 (27)	3 (43)	6 (35)	6 (36)
12-48h	5 (9)	1 (7)	4 (13)	4 (57)	1 (6)	4 (25)
>48h	2 (4)	0 (0)	2 (7)	0 (0)	0 (0)	1 (6)
Absence	28 (52)	6 (43)	16 (53)	4 (57)	10 (59)	5 (31)
Seasonal flare-up, n (%)						
Yes	11 (20)	5 (36)	4 (13)	1 (14)	6 (35)	2 (13)
No	43 (80)	9 (64)	26 (87)	6 (86)	11 (65)	14 (88)
Trigger factor, n (%)						
Cold	24 (44)	11 (79)	11 (37)	2 (29)	11 (65)	7 (44)
Change of temperature	13 (24)	5 (36)	5 (17)	2 (29)	8 (47)	4 (25)
Physical trauma	3 (6)	0 (0)	3 (10)	0 (0)	2 (12)	1 (6)
Fatigue	18 (33)	3 (21)	14 (47)	1 (14)	5 (29)	9 (56)

Supplementary Table S4. Quality of life of all patients, main subtypes and main variants.

	All patients (n=54)	FCAS (n=14)	MWS (n=30)	CINCA (n=7)	A439V (n=17)	R260W (n=16)
Health						
State of health, n (%)						
Excellent	4 (7)	2 (14)	2 (7)	0 (0)	3 (18)	1 (6)
Very good	8 (15)	1 (7)	6 (20)	0 (0)	1 (6)	2 (13)
Good	34 (63)	9 (64)	19 (63)	5 (71)	11 (65)	11 (69)
Poor	6 (11)	1 (7)	2 (7)	2 (29)	1 (6)	1 (6)
Bad	2 (4)	1 (7)	1 (3)	0 (0)	1 (6)	1 (6)
Emotional state, n (%)						
Excellent	5 (9)	1 (7)	3 (10)	1 (14)	1 (6)	2 (13)
Very good	7 (13)	2 (14)	4 (13)	0 (0)	3 (18)	1 (6)
Good	29 (54)	8 (57)	16 (53)	4 (57)	9 (53)	9 (56)
Poor	12 (22)	2 (14)	7 (23)	2 (29)	3 (18)	4 (25)
Bad	1 (2)	1 (7)	0 (0)	0 (0)	1 (6)	0 (0)
Tobacco, n (%)						
Currently	9 (17)	3 (21)	5 (17)	1 (14)	4 (24)	4 (25)
Stopped	15 (28)	5 (36)	8 (27)	1 (14)	5 (29)	4 (25)
Never	30 (56)	6 (43)	17 (57)	5 (71)	8 (47)	8 (50)
Activities						
Physical activity, n (%)						
Very often	3 (6)	0 (0)	2 (7)	1 (14)	2 (12)	0 (0)
Often	17 (32)	5 (36)	11 (37)	0 (0)	6 (35)	5 (31)
Sometimes	12 (22)	0 (0)	8 (27)	3 (43)	1 (6)	6 (38)
Rarely	16 (30)	8 (58)	5 (17)	2 (29)	7 (41)	2 (13)
Never	6 (11)	1 (7)	4 (13)	1 (14)	1 (6)	3 (19)
Indoor/Outdoor						
Indoor	14 (34)	5 (63)	4 (16)	3 (60)	5 (42)	2 (15)
Outdoor	27 (66)	3 (38)	21 (84)	2 (40)	7 (58)	11 (85)

Supplementary Table S4. Quality of life of all patients, main subtypes and main variants. (*Continued*)

	All patients (n=54)	FCAS (n=14)	MWS (n=30)	CINCA (n=7)	A439V (n=17)	R260W (n=16)
Cultural activities, n (%)						
Very often	2 (4)	0 (0)	2 (7)	0 (0)	0 (0)	1 (6)
Often	12 (22)	4 (29)	5 (17)	2 (29)	4 (24)	1 (6)
Sometimes	16 (30)	4 (29)	11 (37)	0 (0)	5 (29)	5 (31)
Rarely	12 (22)	1 (7)	6 (20)	4 (57)	2 (12)	4 (25)
Never	12 (22)	5 (36)	6 (20)	1 (14)	6 (35)	5 (31)
Studies						
School absenteeism, n (%)						
Very often	13 (24)	3 (21)	4 (13)	5 (71)	5 (29)	0 (0)
Often	16 (30)	7 (50)	7 (23)	1 (14)	7 (41)	5 (31)
Sometimes	12 (22)	2 (14)	10 (33)	0 (0)	2 (12)	5 (31)
Rarely	6 (11)	2 (14)	4 (13)	0 (0)	1 (6)	4 (25)
Never	7 (13)	0 (0)	5 (17)	1 (14)	2 (12)	2 (13)
Finished studies, n (%)						
Yes	46 (85)	13 (93)	27 (90)	5 (71)	16 (94)	13 (81)
No	8 (15)	1 (7)	3 (10)	2 (29)	1 (6)	3 (19)
Highest level of education, n (%)						
General educational development	17 (32)	7 (50)	8 (27)	2 (29)	8 (47)	6 (38)
High school diploma	15 (28)	4 (29)	7 (23)	2 (29)	5 (29)	4 (25)
Bachelor's degree	4 (7)	1 (7)	2 (7)	0 (0)	1 (6)	2 (13)
Master's degree	2 (4)	0 (0)	2 (7)	0 (0)	0 (0)	0 (0)
Doctorate	5 (9)	1 (7)	4 (13)	0 (0)	1 (6)	0 (0)
No study	11 (20)	1 (7)	7 (23)	3 (43)	2 (12)	4 (25)
Professional						
Paid professional activity, n (%)						
Yes	28 (52)	8 (57)	16 (53)	3 (43)	9 (53)	7 (44)
No	26 (48)	6 (43)	14 (47)	4 (57)	8 (47)	9 (56)
Disabled worker, n (%)						
< 50%	9 (17)	1 (7)	7 (23)	1 (14)	3 (18)	3 (19)
50-80%	4 (7)	1 (7)	2 (7)	1 (14)	1 (6)	2 (13)
>80%	8 (15)	0 (0)	3 (10)	5 (71)	1 (6)	0 (0)
No disability	33 (61)	12 (86)	18 (60)	0 (0)	12 (71)	11 (69)
Gave up a job, n (%)						
Yes	18 (34)	3 (23)	10 (33)	5 (71)	7 (41)	3 (19)
No	35 (66)	10 (77)	20 (67)	2 (29)	10 (59)	13 (81)
Absenteeism at work, n (%)						
Very often	4 (8)	0 (0)	3 (10)	1 (14)	0 (0)	1 (7)
Often	6 (12)	2 (14)	2 (7)	2 (29)	4 (24)	1 (7)
Sometimes	14 (28)	5 (36)	6 (7)	2 (29)	5 (29)	4 (27)
Rarely	7 (14)	3 (21)	3 (10)	1 (14)	3 (18)	1 (7)
Never	20 (39)	4 (29)	15 (52)	1 (14)	5 (29)	8 (53)
Social relationship						
Live, n (%)						
Alone	3 (6)	0 (0)	0 (0)	3 (43)	2 (12)	0 (0)
With a partner	34 (63)	13 (93)	21 (70)	0 (0)	13 (77)	10 (63)
With a parent	14 (26)	0 (0)	8 (27)	4 (57)	1 (6)	5 (31)
Other	3 (6)	1 (7)	1 (3)	0 (0)	1 (6)	1 (6)
Sentimental relationship, n (%)						
Yes	37 (69)	14 (100)	21 (70)	2 (29)	16 (94)	10 (63)
No	17 (32)	0 (0)	9 (30)	5 (71)	1 (6)	6 (38)
Friendly relationship, n (%)						
Yes	49 (91)	14 (100)	27 (90)	5 (71)	16 (94)	14 (88)
No	5 (9)	0 (0)	3 (10)	2 (29)	1 (6)	2 (13)

Supplementary Table S5. SF36 score of all patients, main subtypes and main variants.

Dimension Médiane (Quartile)	General French population	All patients (n=54)	FCAS (n=14)	MWS (n=30)	CINCA (n=7)
PF	95 (100-80)	85 (100-55)	88 (100-50)	93 (100-65)	65 (75-45)
RP	100 (100-75)	100 (100-50)	100 (100-50)	100 (100-50)	50 (75-50)
BP	74 (100-52)	74 (100-55)	69 (80-45)	80 (100-65)	45 (100-33)
GH	72 (82-57)	50 (65-35)	48 (60-35)	50 (65-40)	40 (60-25)
VT	60 (75-50)	50 (55-35)	45 (50-35)	50 (60-45)	50 (65-25)
SF	88 (100-63)	81 (88-50)	75 (88-50)	81 (100-63)	63 (88-50)
RE	100 (100-67)	100 (100-67)	100 (100-33)	100 (100-67)	100 (100-67)
MH	72 (80-60)	64 (80-48)	60 (64-52)	64 (80-48)	44 (80-20)

PF: physical function; RP: role physical; BP: bodily pain; GH: general health; VT: vitality; SF: social function; RE: role emotional; MH: mental health.

Supplementary Table S6. SF36 score of main variants.

Dimension Mean (Quartile)	A439V (n=17)	R260W (n=16)
PF	90 (100-50)	75 (100-53)
RP	100 (100-50)	100 (100-38)
BP	70 (80-45)	69 (90-55)
GH	55 (65-40)	45 (60-33)
VT	50 (55-45)	50 (55-35)
SF	88 (88-63)	63 (100-57)
RE	100 (100-67)	100 (100-67)
MH	60 (64-62)	64 (71-48)

PF: physical function; RP: role physical; BP: bodily pain; GH: general health; VT: vitality; SF: social function; RE: role emotional; MH: mental health.