

**Supplementary Table S1.** Genetic results of the 46 patients with monogenic autoinflammatory diseases and ocular involvement.

| Monogenic autoinflammatory disease | Gene                   | Genetic variants | N         |
|------------------------------------|------------------------|------------------|-----------|
| <b>CAPS</b>                        | <b><i>NLRP3</i></b>    |                  | <b>13</b> |
|                                    |                        | A439V/WT         | 3         |
|                                    |                        | R260W/WT         | 3         |
|                                    |                        | D303G/WT         | 1         |
|                                    |                        | H49R/WT          | 1         |
|                                    |                        | R262W/WT         | 1         |
|                                    |                        | V198M/WT         | 1         |
|                                    |                        | V200M/WT         | 1         |
|                                    |                        | D303H/WT         | 1         |
|                                    |                        | Q703K/WT         | 1         |
| <b>FMF</b>                         | <b><i>MEFV</i></b>     |                  | <b>12</b> |
|                                    |                        | I591T//WT        | 2         |
|                                    |                        | E148Q/WT         | 2         |
|                                    |                        | P369S-R408Q/WT   | 2         |
|                                    |                        | M594V/M694I      | 1         |
|                                    |                        | R348H/I591T      | 1         |
|                                    |                        | M694V/WT         | 1         |
|                                    |                        | M694I/WT         | 1         |
|                                    |                        | A744S/WT         | 1         |
| K695R/WT                           | 1                      |                  |           |
| <b>TRAPS</b>                       | <b><i>TNFRSF1A</i></b> |                  | <b>9</b>  |
|                                    |                        | R92Q/WT          | 8         |
| <b>HIDS</b>                        | <b><i>MVK</i></b>      | R92Q/R92Q        | 1         |
|                                    |                        |                  | <b>2</b>  |
| <b>BLAU syndrome</b>               | <b><i>NOD2</i></b>     | V377I/WT         | 1         |
|                                    |                        | V377I/WT*        | 1         |
| <b>DADA2</b>                       | <b><i>ADA2</i></b>     |                  | <b>8</b>  |
|                                    |                        | R587C/WT         | 4         |
|                                    |                        | R334Q/WT         | 1         |
|                                    |                        | R334W/WT         | 1         |
|                                    |                        | C495Y/WT         | 1         |
| <b>NLRC4-AD</b>                    | <b><i>NLRC4</i></b>    | L349F/WT         | 1         |
|                                    |                        |                  | <b>1</b>  |
|                                    | <b><i>ADA2</i></b>     | G47R/W264S       | <b>1</b>  |
|                                    | <b><i>NLRC4</i></b>    | T337S/WT         | <b>1</b>  |

CAPS: cryopyrin-associated autoinflammatory syndromes; DADA2: deficiency of adenosine deaminase 2; FMF: familial Mediterranean fever; HIDS: hyperimmunoglobulin D syndrome; NLRC4-AD: NLRC4-associated disease; TRAPS: tumour necrosis factor receptor-associated periodic syndrome; WT: wild type.

\*This patient also had the variant R135H/WT in *NLRP3* gene.

**Supplementary Table S2.** Type of ocular lesions according to the age of disease onset.

|                           | Paediatric onset | Adult onset | Total     | <i>p</i> value* |
|---------------------------|------------------|-------------|-----------|-----------------|
| <b>Number of patients</b> | 25               | 21          | 46        |                 |
| <b>Conjunctivitis</b>     | 10 (40)          | 16 (76.2)   | 26 (56.5) | 0.013           |
| <b>Uveitis (any)</b>      | 18 (72)          | 5 (23.8)    | 23 (50)   | 0.001           |
| <i>Anterior uveitis</i>   | 12 (48)          | 5 (23.8)    | 17 (37)   | 0.011           |
| <i>Panuveitis</i>         | 6 (24)           | 0           | 6 (13)    | 0.009           |
| <b>Keratitis</b>          | 4 (16)           | 4 (19)      | 8 (17.4)  | 0.786           |
| <b>Episcleritis</b>       | 3 (12)           | 4 (19)      | 7 (15.2)  | 0.057           |
| <b>Periorbital oedema</b> | 0                | 2 (9.5)     | 2 (4.3)   | 0.115           |
| <b>Optic disk oedema</b>  | 0                | 1 (4.8)     | 1 (2.2)   | 0.270           |

\*Differences calculated between paediatric and adult patients.

**Supplementary Table S3.** Systemic treatments received at the time of data collection.

| Monogenic autoinflammatory disease | N (%)           | No treatment   | Colchicine (alone) | Prednisone (alone) | Conventional immunosuppressive drugs <sup>a</sup> | Biologic agents            |                           |                          |                          |
|------------------------------------|-----------------|----------------|--------------------|--------------------|---|----------------------------|---------------------------|--------------------------|--------------------------|
|                                    |                 |                |                    |                    |   | IL-1 blockers <sup>b</sup> | TNF blockers <sup>c</sup> | Tocilizumab <sup>d</sup> | Tofacitinib <sup>e</sup> |
| CAPS                               | 13 (28.3)       | 2 (15.3)       | 0                  | 0                  | 0   | 10 (77)                    | 1 (7.6)                   | 0                        | 0                        |
| FCAS                               | 1 (2.2)         | 0              | 0                  | 0                  | 0   | 1 (100)                    | 0                         | 0                        | 0                        |
| MWS                                | 11 (24)         | 2 (18)         | 0                  | 0                  | 0   | 8 (72)                     | 1(9)                      | 0                        | 0                        |
| CINCA-NOMID                        | 1 (2.2)         | 0              | 0                  | 0                  | 0   | 1 (100)                    | 0                         | 0                        | 0                        |
| FMF                                | 12 (26)         | 2 (16.7)       | 6 (50)             | 0                  | 0   | 3 (25)                     | 0                         | 1 (8)                    | 0                        |
| TRAPS                              | 9 (20)          | 0              | 1 (11)             | 0                  | 2 (22)  | 6 (66.7)                   | 0                         | 0                        | 0                        |
| BLAU syndrome                      | 8 (17)          | 0              | 0                  | 1 (12.5)           | 1 (12.5)  | 0                          | 5 (62.5)                  | 0                        | 1 (12.5)                 |
| HIDS                               | 2 (4.4)         | 0              | 0                  | 0                  | 0   | 2 (100)                    | 0                         | 0                        | 0                        |
| DADA2                              | 1 (2.2)         | 0              | 0                  | 0                  | 0   | 0                          | 1 (100)                   | 0                        | 0                        |
| NLRC4-AD                           | 1 (2.2)         | 0              | 0                  | 0                  | 0   | 1 (100)                    | 0                         | 0                        | 0                        |
| <b>TOTAL</b>                       | <b>46 (100)</b> | <b>4 (8.7)</b> | <b>7 (15.2)</b>    | <b>1 (2.2)</b>     | <b>3 (6.5)</b>                                    | <b>22 (47.8)</b>           | <b>8 (17.4)</b>           | <b>1 (2.2)</b>           | <b>1 (2.2)</b>           |

<sup>a</sup>Methotrexate (n=1), Prednisone/Methotrexate (n=1), Prednisone/Colchicine/Methotrexate (n=1).

<sup>b</sup>Anakinra (n=14), canakinumab (n=8).

<sup>c</sup>Adalimumab (n=3; 1 with prednisone), Etanercept (n=1), Certolizumab (n=1), Methotrexate/Adalimumab (n=1), Prednisone/Tacrolimus/Adalimumab (n=1).

<sup>d</sup>Tocilizumab with colchicine (n=1).

<sup>e</sup>Azathioprine/Tofacitinib (n=1).