|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **cDNA position** | **Protein domain** | **Previous reference** | **M/F** | **Age (years)** | **CMC** | **Other infections** | **Auto**  **immunity** | **Auto**  **antibodies** | **Pneumopathy** | **Aneurysms/ malignancy** | **Others** | **Treatment** |
| **P1E29A**  **Mašek** | c.86A>C | N-terminal | 1 | M | 45 | **+** | SPI | AIHA, TH, AIH, CD | Coombs, a-TTG, a-EM,a-TG | BE | -/- | AS, KT | AZ, CS, RTX, RX, |
| **P2Y68C**  **Poešánová J** | c.203A>G | N-terminal | 1 | F | 21 | **+** | SPI | - | ASCA | - | -/- | AS, KT, LFT elevation, VC | AZ, RX, IgRT |
| **P3Y68C**  **Pešánová M** | c.203A>G | N-terminal | 1 | F | 45 | **+** | SPI | TH | ASCA, ANA, a-GLD | BA | -/- | AS, KT, LFT elevation, VC | AZ |
| **P4A267V**  **Potužníková J** | c.800C>T | Coiled-coil | 2 | F | 46 | **+** | SPI  HSV | TH | ASCA | - | -/- | KT, CG, OeS, VC | AZ, TMP |
| **P5**T288N  **Čubanová** | c.863C>A | Coiled-coil | 1 | F | 52 | + | SPI  HSV | - | ASCA, CCP, AECA | MVI | Aortal aneurysm/ - | AS, KT, LFT elevation | AZ |
| **P6N357D**  **Melichar** | c.1069A>G | DNA-binding | This report | M | 8 | **+** | OMA | - | - | - | -/- | - | AZ |
| **P7N357D**  **Melichar** | c.1069A>G | DNA-binding | This report | M | 45 | **+** | HSV | - | ASCA, RF | - | -/- | - | AZ |
| **P8M390T**  **Křiváčková** | c.1169T>C | DNA-binding | 1 | F | 18 | **+** | SPI  VZV | - | - | BE, AT, MVI | -/- | AS, KT | AMP, AZ, RX, IgRT |

**Supplementary Table 1: Genetic and clinical characteristics of STAT1 GOF patients.**

a-AECA – anti-endothelial cell antibodies, a-CCP – anti-cyclic citrulinated peptide, a-EM – anti-endomysium, a-GLD – anti-gliadin antibodies, a-TG – anti-thyreoglobulin, a-TTG – anti-thyreoglobulin, AIH – autoimmune hepatitis; AIHA – autoimmune haemolytic anemia; ANA – anti-nuclear antibodies, AS – aphthous stomatitis; ASCA – anti-saccharomyces cerevisiae, AT – atelectases; BA – bronchial asthma; BE – bronchiectasis; CD – celiac disease; CG – chronic gastritis; CMC - chronic mucocutaneous candidiasis; HSV – herpes simplex virus; KT - keratitis; LFT – liver function tests; M/F – male/female‚ MVI - mixed ventilatory impairment; OeS – oesophageal strictures; OMA – otitis media; RF – rheumathoid factor, SPI – sinopulmonary infections; TH – thyroiditis; VC – vaginal candidiasis; VZV – varicella zoster virus;

**Treatment:** AMP – amphotericin B, AZ – azoles, CS – corticosteroids, IgRT – immunoglubulin replacement therapy, RTX – rituximab, RX – ruxolitinib, TMP - trimethoprim

*1Bloomfield et al. Immunogenicity and Safety of COVID-19 mRNA Vaccine in STAT1 GOF Patients. J Clin Immunol. 2021**Oct 31. doi: 10.1007/s10875-021-01163-8*

*2Soltész B, Tóth B, Shabashova N, et al New and recurrent gain-of-function STAT1 mutations in patients with chronic mucocutaneous candidiasis from Eastern and Central Europe Journal of Medical Genetics 2013;50:567-578.*

**Supplementary Table 2: Immunologic characteristics of STAT1 GOF patients**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **IgG\*** | **IgG1\*** | **IgG2\*** | **IgG3\*** | **IgG4\*** | **IgA\*** | **IgM\*** | **T cells°** | **Th17°** | **B cells°** | **Neutrophil count°** | **Monocyte count°** | **CRP** | **pSTAT1° (Tyr701)** |
| **P1E29A** |  |  | **↓** |  | **↓** |  |  |  | **↓** | **↓** |  |  | **↑** | **↑** |
| **P2Y68C** |  |  |  |  |  | **↑** | **↓** |  |  |  |  |  | **↑** | **↑** |
| **P3Y68C** | **↑** | **↑** |  |  | **↑** | **↑** |  |  | **↓** | **↓ CD27+ memory** |  |  | **↑** | **↑** |
| **P4A267V** | **↑** | **↑** |  | **↑** | **↓** | **↓** |  |  | **↓** |  |  |  |  | **↑** |
| **P5T288N** |  |  |  | **↑** | **↓** |  |  | **↓ CD4** | **↓** | **↓ CD27+ memory**  **↓ Class switched** |  |  | **↑** | **↑** |
| **P6N357D** |  |  |  | N | **↓** |  | **↓** |  | **↓** | **↑ IgD-CD27- naive** |  |  |  | **↑** |
| **P7N357D** |  |  |  |  |  | **↑** |  |  | **↓** |  |  |  |  | **↑** |
| **P8M390T** | **↑** | **↑** |  | **↑** | **↓** |  |  | **↓ CD4** | **↓** |  |  |  | **↑** | **↑** |

**\* = prior to immunoglobulin replacement therapy, where applicable; ° = prior to ruxolitinib, where applicable; Th17 = CD4+ IL17+; pSTAT1 = IFNγ/ IFNα -induced phosphorylation of p-STAT1 (Tyr701) in CD3+ T lymphocytes; CRP = C-reactive protein**

**Normal, Abnormal, ↑ increased, ↓ decreased**