**Supplementary table**

**Supplementary Table 1.** Documented pathogens associated with STAT1 GOF mutations

|  |  |
| --- | --- |
| **Pathogen** | **Results (%)** |
| Mucocutaneous fungal infections Candida albicans DematophytesFusarium solani OthersInvasive fungal infections Candida albicans Pneumocystis jirovecii Cryptococcus Aspergillus Penicillium marneffei OthersBacterial infections Staphylococcus aureus Streptococcus spp. Pseudomonas aeruginosa Haemophilus influenzae Klebsiella pneumoniaSalmonella Clostridium difficileOthersMycobacterial infections M tuberculosisBCG strainOthersViral infections Herpes simplex Varicella-zosterCMV or EBV Molluscum contagiosum/wartsEncephalitis B virus JCV Others | n=244n=209 (85.7)n=26 (10.7)n=1 (0.4)n=10 (4.1)n=55n=10 (18.2)n=11 (20.0)n=12 (21.8)n=11 (20.0)n=5 (9.1)n=8 (14.5)n=138n=55 (39.9)n=32 (23.2)n=22 (15.9)n=16 (11.6)n=1 (0.7)n=1 (0.7)n=2 (1.4)n=25 (18.1)n=40n=18 (45.0)n=12 (30.0)n=13 (32.5)n=216n=52 (24.1)n=73 (33.8)n=47 (21.8)n=44 (20.4)n=1 (0.5)n=4 (1.9)n=25 (11.6) |
| Parasitic infections Toxoplasma gondiiDemodexLeishmania | n=9n=1 (11.1)n=5 (55.6)n=2 (22.2) |

**Supplementary Table 2.** Documented pathogens associated with STAT1 LOF mutations

|  |  |
| --- | --- |
| **Pathogens** | **Results (%)** |
| Mycobacterial infections M. tuberculosisBCG strainM. aviumM. szulgaiM. kansasiiNot knownViral infections Herpes simplex Varicella-zoster CMV EBV Molluscum contagiosum/warts HHV RSVAdenovirusParvovirusBacterial infectionsSalmonella infection | n=28n=2 (7.1)n=17 (60.7)n=5 (17.9)n=1 (3.6)n=1 (3.6)n=3 (10.7)n=12n=5 (41.7)n=5 (41.7)n=5 (41.7)n=6 (50.0)n=2 (16.7)n=4 (33.3)n=4 (33.3)n=3 (25.0)n=1 (8.3)n=5n=2 (40.0) |
| ShigellaKlebsiellaE. coliStaphylococcusInvasive fungal infections Candida albicans AspergillusParasitic infections Toxoplasma gondii (IgG) | n=1 (20.0)n=1 (20.0)n=1 (20.0)n=1 (20.0)n=2n=1 (50.0)n=1 (50.0)n=1n=1 (100.0) |

**Supplementary Table 3.** Site of infection in patients with STAT1 GOF mutations

|  |  |
| --- | --- |
| **Type of infection** | **Patients (%) (n=442)** |
| Mucocutaneous fungal infections Oropharyngeal mycosis Cutaneous mycosis Esophageal/genital mycosis Onychomycosis Aphtous stomatitis Scalp mycosisInvasive fungal infections Invasive candidiasis Other invasive infectionsBacterial infectionsLRIENTSkinOthersMycobacterial infections Lung disease Adenitis/skin diseaseDisseminated diseaseViral infections Cutaneous Systemic | n=363 (82.1)n=335 (75.8)n=191 (43.2)n=186 (42.1)n=189 (42.8)n=138 (31.2)n=56 (12.7)n=49 (11.1)n=15 (3.4)n=36 (8.1)n=257 (58.1)n=164 (37.1)n=150 (33.9)n=97 (21.9)n=33 (7.5)n=41 (9.3)n=16 (3.6)n=15 (3.4)n=13 (2.9)n=130 (29.4)n=108 (24.4)n=35 (7.9) |

**Supplementary Table 4.** Sites of infection in patients with STAT1 LOF mutations

|  |  |
| --- | --- |
| **Type of infection** | **Results (%)** |
| Mycobacterial infections Lung disease Adenitis/skin diseaseBone diseaseLiver/spleen diseaseBrain diseaseOcular diseaseGastrointestinal diseaseDisseminated diseaseViral infections Cutaneous Brain diseaseLung diseaseLiver disease SystemicBacterial infectionsLRIENTSkinBrainLiverBacteremiaMucocutaneous fungal infectionsInvasive fungal infections | n=24n=4 (16.7)n=14 (58.3)n=15 (62.5)n=4 (16.7)n=2 (8.3)n=1 (4.2)n=1 (4.2)n=19 (79.2)n=7n=4 (57.1)n=3 (42.9)n=2 (28.6)n=1 (14.3)n=7 (100)n=5n=1 (20.0)n=3 (60.0)n=1 (20.0)n=1 (20.0)n=1 (20.0)n=3 (60.0)n=1n=2 |

**Supplementary Table 5.** Immunological investigation of patients with the STAT1 GOF mutation

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Biological investigation** | **Patients (n)** | **Normal (%)** | **Low (%)** | **High (%)** |
| Total lymphocytes | 291 | 75.6 | 23.0 | 1.4 |
| CD4+ T lymphocytes | 276 | 68.1 | 30.8 | 1.1 |
| CD8+ T lymphocytes | 268 | 79.5 | 17.9 | 2.6 |
| CD19+ or CD20+ B lymphocytes | 264 | 74.2 | 23.9 | 1.9 |
| CD27+CD19+ memory B lymphocytes | 63 | 47.6 | 50.8 | 1.6 |
| CD16+CD56+ natural killer cells | 209 | 66.5 | 32.1 | 1.4 |
| CD3+IL-17+ or CD4+IL-17+ T lymphocytes | 90 | 12.2 | 87.8 | 0 |
| T cell proliferation (mitogen and/or antigen) | 149 | 67.8 | 32.2 | 0 |
| IgG levels (g/L) | 294 | 73.1 | 6.5 | 20.4 |
| IgA levels (g/L) | 273 | 75.8 | 18.3 | 6.2 |
| IgM levels (g/L) | 272 | 93.0 | 6.3 | 0.7 |
| IgG1 levels (g/L) | 102 | 99.0 | 1.0 | 0 |
| IgG2 levels (g/L) | 107 | 60.7 | 39.3 | 0 |
| IgG4 levels (g/L) | 105 | 52.4 | 47.6 | 0 |
| IgE levels (g/L) | 162 | 94.4 | 1.2 | 4.3 |
| Antibodies against protein antigens | 134 | 72.4 | 27.6 | 0 |

**Supplementary Table 6.** Immunological investigation of patients with the STAT1 LOF mutation

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Biological investigation** | **Patients (n)** | **Normal (%)** | **Low (%)** | **High (%)** |
| WBC | 15 | 73.3 | 0.0 | 26.7 |
| Neutrophils | 13 | 92.3 | 0.0 | 7.7 |
| Monocyte | 4 | 100.0 | 0.0 | 0.0 |
| Eosinophils | 6 | 66.7 | 0.0 | 33.3 |
| Total lymphocyte | 14 | 92.9 | 0.0 | 7.1 |
| CD4+ T lymphocytes | 12 | 100.0 | 0.0 | 0.0 |
| CD8+ T lymphocytes | 12 | 91.7 | 0.0 | 8.3 |
| CD19+ or CD20+ B lymphocytes | 12 | 100.0 | 0.0 | 0.0 |
| CD27+CD19+ memory B lymphocytes | 2 | 100.0 | 0.0 | 0.0 |
| CD16+CD56+ natural killer cells | 5 | 80.0 | 0.0 | 20.0 |
| T cell proliferation (mitogen and/or antigen) | 3 | 100.0 | 0.0 | 0.0 |
| IgG levels (g/L) | 12 | 83.3 | 0.0 | 16.7 |
| IgA levels (g/L) | 10 | 100.0 | 0.0 | 0.0 |
| IgM levels (g/L) | 10 | 100.0 | 0.0 | 0.0 |
| IgE levels (g/L) | 10 | 100.0 | 0.0 | 0.0 |
| Antibodies against protein antigens | 4 | 75.0 | 25.0 | 0.0 |

WBC, White blood cell.

**Supplementary Figure**

**Supplementary Figure 1.** Kaplan–Meier curves. (A) Onset of CMC for patients with GOF. (B) Overall survival curves for patients with GOF. (C) Overall survival curves for patients with LOF.



Figure 1A.



Figure 1B.



Figure 1C.

**Supplementary Figure 2.** Several strains of mycobacteria.



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