**Supplementary table**

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

|  |  |
| --- | --- |
| **Pathogen** | **Results (%)** |
| Mucocutaneous fungal infections  Candida albicans  Dematophytes  Fusarium solani  Others  Invasive fungal infections  Candida albicans  Pneumocystis jirovecii  Cryptococcus  Aspergillus  Penicillium marneffei  Others  Bacterial infections  Staphylococcus aureus  Streptococcus spp.  Pseudomonas aeruginosa  Haemophilus influenzae  Klebsiella pneumonia  Salmonella  Clostridium difficile  Others  Mycobacterial infections  M tuberculosis  BCG strain  Others  Viral infections  Herpes simplex  Varicella-zoster  CMV or EBV  Molluscum contagiosum/warts  Encephalitis B virus  JCV  Others | n=244  n=209 (85.7)  n=26 (10.7)  n=1 (0.4)  n=10 (4.1)  n=55  n=10 (18.2)  n=11 (20.0)  n=12 (21.8)  n=11 (20.0)  n=5 (9.1)  n=8 (14.5)  n=138  n=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=40  n=18 (45.0)  n=12 (30.0)  n=13 (32.5)  n=216  n=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 gondii  Demodex  Leishmania | n=9  n=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. tuberculosis  BCG strain  M. avium  M. szulgai  M. kansasii  Not known  Viral infections  Herpes simplex  Varicella-zoster  CMV  EBV  Molluscum contagiosum/warts  HHV  RSV  Adenovirus  Parvovirus  Bacterial infections  Salmonella infection | n=28  n=2 (7.1)  n=17 (60.7)  n=5 (17.9)  n=1 (3.6)  n=1 (3.6)  n=3 (10.7)  n=12  n=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=5  n=2 (40.0) |
| Shigella  Klebsiella  E. coli  Staphylococcus  Invasive fungal infections  Candida albicans  Aspergillus  Parasitic infections  Toxoplasma gondii (IgG) | n=1 (20.0)  n=1 (20.0)  n=1 (20.0)  n=1 (20.0)  n=2  n=1 (50.0)  n=1 (50.0)  n=1  n=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 mycosis  Invasive fungal infections  Invasive candidiasis  Other invasive infections  Bacterial infections  LRI  ENT  Skin  Others  Mycobacterial infections  Lung disease  Adenitis/skin disease  Disseminated disease  Viral 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 disease  Bone disease  Liver/spleen disease  Brain disease  Ocular disease  Gastrointestinal disease  Disseminated disease  Viral infections  Cutaneous  Brain disease  Lung disease  Liver disease  Systemic  Bacterial infections  LRI  ENT  Skin  Brain  Liver  Bacteremia  Mucocutaneous fungal infections  Invasive fungal infections | n=24  n=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=7  n=4 (57.1)  n=3 (42.9)  n=2 (28.6)  n=1 (14.3)  n=7 (100)  n=5  n=1 (20.0)  n=3 (60.0)  n=1 (20.0)  n=1 (20.0)  n=1 (20.0)  n=3 (60.0)  n=1  n=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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