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Supplementary

**Supplementary Table 1.** Summary of immunological phenotype categories and causal genes identified in this cohort

| Patient no | IUIS (Union of Immunological Societies) 2024 Phenotype Category | Gene | Variant |
|---|---|---|---|
| 1 | Autoimmune lymphoproliferative syndrome (ALPS) | *FAS* NM_000043.6 | c.869C>T p.(Ala290Val) |
| 2 | Immune dysregulation | *LRBA* NM_001364905.1 | c.6372del p.(F2124Lfs*29) |
| 3 | T-B- combined immune deficiency | *RAG2* NM_000536.4 | c.233G>C p.(Cys78Ser) |
| 4 | Combined immunodeficiency | *CIITA* NM_000246.4 | c.2879T>A p.(Leu960Gln) |
| 5 | T-B- severe combined immune deficiency | *DCLRE1C* | Exon1-3 deletion[&] |
| 6 | T-B- severe combined immune deficiency | *DCLRE1C* NM_001033855.3 | c.632G>T p.(Gly211Val) |
| 7 | Mendelian susceptibility to mycobacterial disease | *STAT1* NM_007315.4 | c.1154C>T p.(Thr385Met) |
| 8 | Mendelian susceptibility to mycobacterial disease | *STAT1* NM_007315.4 | c.71A>G p.(Asp24Gly) |
| 9 | Agammaglobulinemia | *BTK* NM_000061.3 | c.900_903del p.(Gly302Valfs*28) |
| 10 | T-B- severe combined immune deficiency | *RAG1* NM_000448.3 | c.1682G>A p.(Arg561His) |
| 11 | T-B- severe combined immune deficiency | *RAG1* NM_000448.3 | c.1767C>G p.(Tyr589*) |
| 12 | Immuno-osseous dysplasia | *SMARCAL1* NM_014140.4 | c.1939A>C p.(Lys647Gln) |
| 13 | Mendelian susceptibility to mycobacterial disease | *IFNGR1* NM_000416.3 | c.523del p.(Tyr175Metfs*2) |
| 14 | Syndromes associated with elevated IgE and/or atopic disease | *SPINK5* NM_006846.4 | c.238dup p.(Ala80Glyfs*19) and c.1888-1G>A |
| 15 | Syndromes associated with elevated IgE and/or atopic disease | *SPINK5* NM_006846.4 | c.1351dup p.(Cys451Leufs*6) |
| 16 | Regulatory T-cell defects | *FOXP3* NM_014009.4 | c.1040G>A p.(Arg347His) |
| 17 | Combined immunodeficiencies with associated or syndromic features | *TTC37* NM_014639.4 | c.66C>G, p.(Tyr22*) |
| 18 | Autoimmunity with or without lymphoproliferation | *AIRE* *NM_000383.4* | c.769C>T p.(Arg257*) |
| 19 | Autoinflammatory Disorders | *LPIN2* NM_001375808.2 | c.1673G>A p.(Trp558*) |
| 20 | Thymic defects with additional congenital anomalies | *FOXN1* NM_001369369.1 | c.880G>A p.(Val294Ile) |

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**Supplementary Table 1.** Continued

| Patient no | IUIS (Union of Immunological Societies) 2024 Phenotype Category | Gene | Variant |
|---|---|---|---|
| 21 | Combined immunodeficiency | *MALT1*<br>NM_006785.4 | c.1202_1203insAAT<br>p.(Leu401_Leu402insIle) |
| 22 | Anhidrotic ectodermodysplasia with immunodeficiency | *IKBKG*<br>NM_001099857.5 | c.64del<br>p.(Ala22Glnfs*93) |
| 23 | Combined immunodeficiency | *DOCK8* | Exon2-26 deletion[&] |
| 24 | Severe reduction in at least 2 serum immunoglobulin isotypes with normal or low number of B cells, combined immunodeficiency phenotype | *NFKB2*<br>NM_001322934.2 | c.2557C>T<br>p.(Arg853*) |
| 25 | Mendelian susceptibility to mycobacterial disease | *TYK2*<br>NM_003331.5 | c.647del<br>p.(Pro216Argfs*14) |

**List of the genes**

*ABCB1 ACP5 ACTB ADA ADAM17 ADAR AICDA AIRE AK2 AP3B1 AP3D1 APOL1 ATG16L1 ATM B2M BCL10 BLM BLNK BLOC1S6 BTK C1QA C1QB C1QC C1R C1S C3 C5 C6 C7 C8A C8B C9 CARD11 CARD14 CARD9 CASP10 CASP8 CCBE1 CCL2 CD19 CD247 CD27 CD3D CD3E CD3G CD40 CD40LG CD46 CD55 CD59 CD79A CD79B CD81 CD8A CEBPE CFD CFH CFHR1 CFHR5 CFI CFP CHD7 CIITA CLEC7A CORO1A CR2 CSF2RA CSF2RB CSF3R CTC1 CTLA4 CTSC CXCR4 CYBA CYBB DCLRE1C DGKE DKC1 DNMT3B DOCK8 EGFR ELANE EPCAM EXTL3 F12 FADD FAS FASLG FCN3 FERMT3 FOXN1 FOXP3 FPR1 G6PC3 G6PD GATA2 GFI1 GUCY2C HAX1 HMOX1 ICOS IFIH1 IFNGR1 IFNGR2 IGLL1 IKBKB IKBKG IKZF1 IL10 IL10RA IL10RB IL12B IL12RB1 IL17F IL17RA IL21R IL23R IL2RA IL2RG IL7R IRAK4 IRF5 IRF8 IRGM ITCH ITGAM ITGB2 ITK JAK3 KDM6A KMT2D KRAS LAMTOR2 LCK LIG1 LIG4 LPIN2 LRBA LYST MAGT1 MALT1 MAP3K14 MASP1 MASP2 MBL2 MC2R MC3R MCM4 MKL1 MOGS MPO MRAP MRC1 MRE11 MRTFA MS4A1 MSH6 MST1 MTHFD1 MYD88 MYO5B NBN NCF2 NCF4 NEUROG3 NFKB2 NFKBIA NHEJ1 NOD2 NOP10 NRAS ORAI1 PHF11 PIK3CD PIK3R1 PLCG2 PMS2 PNP POLD1 POLE2 PRF1 PRKDC PRPS1 PTEN PTPN22 PTPRC RAB27A RAC2 RAG1 RAG2 RASGRP1 REL RFX5 RFXANK RFXAP RHOH RNASEH2A RNASEH2B RNASEH2C RNF168 SAMD9 SAMHD1 SEMA3E SERPING1 SH2D1A SH3BP2 SLC11A1 SLC26A3 SLC35C1 SLC37A4 SLC46A1 SMARCAL1 SP110 SPINK5 SPINT2 STAT1 STAT3 STAT5B STIM1 STK11 STK4 STXBP2 TAP2 TAZ TBK1 TBX1 TCF3 TCN2 TERC TERT TFRC THBD TICAM1 TINF2 TIRAP TLR1 TLR2 TLR3 TMC6 TMC8 TMEM173 TNFRSF13B TNFRSF13C TRAF3 TRAF3IP2 TREX1 TTC37 TYK2 UNC119 UNC13D UNC93B1 UNG USB1 VPS13B WAS WIPF1 WRAP53 XIAP ZAP70 ZBTB24*