

Gener för medfödda immundefekter (n=442)

| | | |
|-----------|----------|---------|
| BTK | TRNT1 | IL10 |
| IGHM | CLEC16A | PSTPIP1 |
| IGLL1 | IL21 | NOD2 |
| CD79A | NOTCH1 | LPIN2 |
| CD79B | RTP4 | IL1RN |
| BLNK | MAP3K14 | HOIL1 |
| LRRC8A | PMS2 | IL36RN |
| ICOS | TTC37 | SLC29A3 |
| CD19 | INO80 | CARD14 |
| CD81 | MSH6 | SH3BP2 |
| MS4A1 | NFKB1 | PSMB8 |
| TNFRSF13B | IRF2BP2 | PLCG2 |
| TNFRSF13C | ATP6AP1 | CECR1 |
| CR2 | TOP2B | NFAT5 |
| CD40LG | TNFSF10 | NLRC4 |
| CD40 | IRF4 | ADAM17 |
| AICDA | OAS1 | WDR1 |
| UNG | ARHGEF1 | COPA |
| CARD11 | TNFSF13 | NLRP1 |
| IL21R | SLC39A7 | OTULIN |
| LRBA | MEFV | TNFAIP3 |
| MOGS | MVK | DOCK11 |
| TCF3 | NLRP3 | SHARPIN |
| PIK3R1 | NLRP12 | SEC61A1 |
| TNFSF12 | TNFRSF1A | PSMB9 |
| NFKB2 | IL10RA | PSMA3 |
| PRKCD | IL10RB | POMP |

| | | |
|---------|---------|----------|
| PSMB4 | CTLA4 | C3 |
| IRF9 | CD28 | C5 |
| IL18BP | NEIL3 | C6 |
| CDC42 | TFRC | C7 |
| TGFB1 | BCL10 | C8A |
| TAP2 | RELB | C8B |
| TAP1 | GINS1 | C8G |
| TAPBP | RASGRP1 | C9 |
| RFX5 | CD70 | SERPING1 |
| RFXAP | CARMIL2 | CFB |
| MHC2TA | COPG1 | CFD |
| RFXANK | REL | CFP |
| B2M | HAVCR2 | CFI |
| ORAI1 | CHUK | CFH |
| STIM1 | RELA | MBL2 |
| CD8A | TNFRSF9 | MASP1 |
| ZAP70 | POLD1 | MASP2 |
| RHOH | POLD2 | COLEC1 |
| MSN | FCHO1 | CD46 |
| PIK3CD | C1QA | PIGA |
| CTPS1 | C1QB | CD59 |
| IKBKB | C1QC | FCN3 |
| TRAC | C1S | C1QBP |
| MALT1 | C1R | CD55 |
| TNFRSF4 | C4A | CFHR1 |
| ZBTB24 | C4B | CFHR3 |
| DOCK2 | C2 | THBD |

| | | |
|---------|---------|---------|
| TPP2 | CTC1 | RAC2 |
| IKBKG | WRAP53 | ACTB |
| NFKBIA | STN1 | FPR1 |
| IRAK4 | SAMD9 | CTSC |
| MYD88 | SAMD9L | CEBPE |
| CXCR4 | PRF1 | SBDS |
| TMC6 | UNC13D | CYBB |
| TMC8 | STX11 | CYBA |
| STAT2 | STXBP2 | NCF1 |
| RPSA | SH2D1A | NCF2 |
| IRF7 | XIAP | NCF4 |
| IFNAR2 | ITK | GATA1 |
| IRAK1 | TNFRSF7 | CSF3R |
| TIRAP | BLOC1S6 | VPS45 |
| HMOX1 | FAAP24 | JAGN1 |
| NBAS | ELANE | SMARCD2 |
| RANBP2 | GFI1 | HYOU1 |
| IFNAR1 | HAX1 | DNAJC21 |
| DKC1 | G6PC3 | G6PD |
| NOP10 | SLC37A4 | CYBC1 |
| TERC | LAMTOR2 | SRP54 |
| TINF2 | TAZ | CD4 |
| NHP2 | VPS13B | TLR3 |
| RTEL1 | USB1 | UNC93B1 |
| DCLRE1B | ITGB2 | TRAF3 |
| PARN | SLC35C1 | TICAM1 |
| ACD | FERMT3 | TBK1 |

| | | |
|---------|-----------|----------|
| IRF3 | SLC11A1 | IKZF1 |
| IL6R | SPPL2A | STAT5B |
| STAT3 | IL12RB1 | MAGT1 |
| DOCK8 | RORC | WAS |
| TYK2 | STAT1 | WIPF1 |
| PGM3 | FCGR3A | VAV1 |
| ZNF341 | MCM4 | ATM |
| LYST | TNFSF11 | MRE11A |
| MYO5A | TNFRSF11A | NBN |
| RAB27A | TCIRG1 | BLM |
| AP3B1 | OSTM1 | DNMT3B |
| AP3D1 | CLCN7 | RNF168 |
| NKX2-5 | SNX10 | SMARCAL1 |
| MKL1 | PLEKHM1 | SPINK5 |
| PIK3CA | CA2 | SP110 |
| PTEN | GATA2 | CHD7 |
| G6PC | CSF2RA | POLE |
| ALG1 | CLBP | TTC7A |
| TCN2 | CSF2RB | SEMA3E |
| SLC46A1 | CARD9 | EPG5 |
| MTHFD1 | IL17RA | RNF31 |
| IL12B | IL17F | ARPC1B |
| IFNGR1 | IL17RC | CDCA7 |
| IFNGR2 | TRAF3IP2 | HELLS |
| IRF8 | CLEC7A | POLE2 |
| ISG15 | APOL1 | NSMCE3 |
| IL12RB2 | RMRP | ERCC6L2 |

| | | |
|----------|----------|----------|
| MYSM1 | DEF6 | SAMHD1 |
| RNU4ATAC | FOXN1 | ADAR |
| EXTL3 | TBX1 | ACP5 |
| TERT | LAT | IFIH1 |
| LCK | IL2RG | POLA1 |
| PNP | JAK3 | USP18 |
| STK4 | IL7R | DDX58 |
| TNFRSF6 | PTPRC | SKIV2L |
| TNFSF6 | CD3D | USP43 |
| CASP10 | CD3E | ICOSLG |
| CASP8 | CD3G | IL6ST |
| NRAS | CD247 | ERBIN |
| FADD | CORO1A | TGFBR1 |
| AIRE | RAG1 | TGFBR2 |
| FOXP3 | RAG2 | NFE2L2 |
| IL2RA | DCLRE1C | SH3KBP1 |
| ITCH | LIG4 | SLC7A7 |
| KRAS | LIG1 | FERMT1 |
| TMEM173 | PRKDC | IL23R |
| BACH2 | AK2 | CIB1 |
| JAK1 | ADA | POLR3A |
| PEPD | NHEJ1 | POLR3C |
| IL2RB | BCL11B | POLR3F |
| GIMAP6 | TREX1 | DBR1 |
| RIPK1 | RNASEH2A | DNASE2 |
| KMT2D | RNASEH2B | DNASE1L3 |
| GIMAP5 | RNASEH2C | PSMG2 |

| | |
|-----------|---------|
| ALPI | SOCS1 |
| TRIM22 | TET2 |
| RC3H1 | TBX21 |
| CD48 | IFNG |
| SNORA31 | NOS2 |
| GUCY2C | MAPK8 |
| SLC26A3 | LSM11 |
| COL7A1 | RNU7-1 |
| PIK3CG | NCKAP1L |
| CARD8 | UBA1 |
| PAX1 | |
| ATG4A | |
| MAP1LC3B2 | |
| POU2AF1 | |
| EFL1 | |
| HPS1 | |
| HPS4 | |
| HPS6 | |
| NPC1 | |
| SLC9A3 | |
| SLCO2A1 | |
| STXBP3 | |
| TLR2 | |
| LCP2 | |
| MCM10 | |
| FNIP1 | |
| CTNBL1 | |