# Primaire immuundeficienties

Primaire immuundeficiënties volledig panel
429
PID00v21.1

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| ELANE | 100 | 100 |
| CFD | 99.8 | 100 |
| TCF3 | 100 | 100 |
| AP3D1 | 98.7 | 97.9 |
| TICAM1 | 100 | 100 |
| CD70 | 100 | 100 |
| C3 | 100 | 100 |
| VAV1 | 100 | 100 |
| INSR | 98.8 | 99 |
| STXBP2 | 100 | 99.7 |
| TYK2 | 100 | 100 |
| ACP5 | 100 | 100 |
| MAN2B1 | 100 | 99.9 |
| RNASEH2A | 100 | 100 |
| DNASE2 | 100 | 100 |
| FCHO1 | 99.1 | 97.4 |
| JAK3 | 100 | 100 |
| IL12RB1 | 100 | 100 |
| RFXANK | 100 | 100 |
| C19orf40 | 100 | 100 |
| PEPD | 100 | 100 |
| PSENEN | 100 | 100 |
| TGFB1 | 99.2 | 100 |
| CD79A | 99.9 | 100 |
| ARHGEF1 | 100 | 99.9 |
| RELB | 99.2 | 99.5 |
| ERCC2 | 100 | 100 |
| LIG1 | 100 | 99.7 |
| IRF3 | 100 | 100 |
| FPR1 | 100 | 100 |
| NLRP12 | 100 | 100 |
| RBCK1 | 99.9 | 100 |
| THBD | 100 | 100 |
| GINS1 | 100 | 100 |
| DNMT3B | 100 | 99.9 |
| ZNF341 | 99.5 | 99.8 |
| ITCH | 95.9 | 97 |
| SAMHD1 | 100 | 100 |
| ADA | 100 | 98.7 |
| STK4 | 100 | 100 |
| CD40 | 100 | 100 |
| RTEL1 | 100 | 100 |
| IL10RB | 100 | 100 |
| IFNAR2 | 100 | 100 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| IFNAR1 | 99 | 99.5 |
| IFNGR2 | 94.4 | 92.7 |
| ICOSLG | 96.6 | 100 |
| AIRE | 99.9 | 100 |
| ITGB2 | 100 | 99.9 |
| IL17RA | 100 | 100 |
| CECR1 | 100 | 100 |
| USP18 | 94.3 | 94.7 |
| TBX1 | 86.8 | 84.3 |
| IGLL1 | 100 | 100 |
| TCN2 | 100 | 100 |
| HMOX1 | 100 | 100 |
| APOL1 | 100 | 100 |
| NCF4 | 100 | 100 |
| CSF2RB | 100 | 99.9 |
| IL2RB | 100 | 100 |
| RAC2 | 100 | 100 |
| ISG15 | 100 | 100 |
| TNFRSF4 | 93.5 | 100 |
| TNFRSF9 | 100 | 100 |
| PIK3CD | 100 | 100 |
| MASP2 | 100 | 100 |
| C1QA | 100 | 100 |
| C1QC | 100 | 100 |
| C1QB | 100 | 100 |
| FCN3 | 99.9 | 100 |
| LCK | 100 | 100 |
| AK2 | 100 | 99.5 |
| CSF3R | 100 | 100 |
| CTPS1 | 100 | 100 |
| C8A | 100 | 100 |
| C8B | 100 | 100 |
| MYSM1 | 100 | 94.6 |
| MKL1 | 100 | 100 |
| TNFRSF13C | 88.7 | 86.8 |
| CSF2RA | 92.1 | 92 |
| TLR7 | 100 | 100 |
| TLR8 | 100 | 100 |
| PIGA | 100 | 100 |
| SH3KBP1 | 98.6 | 99.4 |
| POLA1 | 98.2 | 99 |
| CYBB | 99.9 | 100 |
| KDM6A | 99.6 | 99.9 |
| UBA1 | 100 | 100 |
| CFP | 100 | 99.6 |
| WAS | 97.1 | 99.8 |
| FOXP3 | 99.8 | 100 |
| MSN | 99.4 | 100 |
| IL2RG | 100 | 100 |
| MAGT1 | 99.9 | 98.9 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| BTK | 99.8 | 100 |
| PRPS1 | 100 | 100 |
| ALG13 | 99.8 | 99.7 |
| XIAP | 99.9 | 97.5 |
| SH2D1A | 99.8 | 100 |
| ELF4 | 100 | 99.3 |
| CD40LG | 100 | 100 |
| IRAK1 | 99 | 98.6 |
| TAZ | 98.4 | 99.9 |
| ATP6AP1 | 99.8 | 100 |
| G6PD | 100 | 99.7 |
| IKBKG | 40.5 | 36.8 |
| SH3BP2 | 91.3 | 91.3 |
| WDR1 | 99.9 | 99.7 |
| DKC1 | 99.9 | 100 |
| JAK1 | 100 | 100 |
| BCL10 | 100 | 100 |
| GFI1 | 99.8 | 100 |
| PTPN22 | 100 | 97.8 |
| DCLRE1B | 100 | 100 |
| NRAS | 100 | 100 |
| FCGR1A | 95.8 | 99.5 |
| VPS45 | 100 | 100 |
| RFX5 | 100 | 100 |
| PSMB4 | 100 | 100 |
| RORC | 100 | 100 |
| HAX1 | 100 | 100 |
| IL6R | 100 | 100 |
| ADAR | 100 | 100 |
| LAMTOR2 | 100 | 100 |
| COPA | 100 | 100 |
| NCSTN | 100 | 100 |
| FCGR2A | 100 | 99.8 |
| FCGR3A | 99.9 | 99 |
| FCGR3B | 95 | 96.9 |
| FCGR2B | 82.7 | 81.4 |
| PBX1 | 100 | 100 |
| CD247 | 100 | 100 |
| FASLG | 100 | 100 |
| RC3H1 | 100 | 100 |
| NCF2 | 100 | 100 |
| IVNS1ABP | 100 | 100 |
| CFH | 100 | 98.4 |
| CFHR3 | 93.9 | 84.7 |
| CFHR1 | 93.9 | 87.4 |
| CFHR5 | 100 | 99.6 |
| PTPRC | 100 | 99.1 |
| IL10 | 100 | 100 |
| CD55 | 96.2 | 99.9 |
| CR2 | 99.8 | 99.3 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| CD46 | 100 | 100 |
| GJC2 | 89.4 | 98.3 |
| IRF2BP2 | 99.3 | 100 |
| LYST | 100 | 99.8 |
| NLRP3 | 100 | 100 |
| ADAM17 | 100 | 99.7 |
| NBAS | 100 | 100 |
| NLRC4 | 100 | 100 |
| TTC7A | 99.9 | 100 |
| MOGS | 100 | 100 |
| CD8A | 99.9 | 99.9 |
| MAL | 100 | 100 |
| ZAP70 | 100 | 100 |
| RANBP2 | 100 | 98.9 |
| IL36RN | 100 | 100 |
| IL1RN | 99.9 | 100 |
| RNU4ATAC | 100 | 0 |
| ERCC3 | 100 | 100 |
| CXCR4 | 100 | 100 |
| IFIH1 | 100 | 100 |
| CDCA7 | 100 | 100 |
| WIPF1 | 100 | 99.7 |
| NFE2L2 | 100 | 100 |
| STAT1 | 100 | 96.8 |
| STAT4 | 100 | 97.5 |
| CASP10 | 100 | 100 |
| CASP8 | 100 | 100 |
| CTLA4 | 100 | 100 |
| ICOS | 100 | 100 |
| SMARCAL1 | 100 | 100 |
| NHEJ1 | 100 | 100 |
| AP1S3 | 99.5 | 99.8 |
| SP110 | 100 | 99.8 |
| ALPI | 100 | 100 |
| TRNT1 | 100 | 91 |
| JAGN1 | 100 | 100 |
| IL17RC | 100 | 100 |
| TOP2B | 99.9 | 98.3 |
| MYD88 | 100 | 100 |
| RPSA | 100 | 100 |
| TREX1 | 100 | 100 |
| PRKCD | 100 | 100 |
| DNASE1L3 | 100 | 100 |
| SEC61A1 | 100 | 100 |
| GATA2 | 100 | 100 |
| PCCB | 100 | 100 |
| DBR1 | 99.8 | 100 |
| TERC | 100 | 0 |
| TFRC | 100 | 99.9 |
| RNF168 | 100 | 100 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| RHOH | 100 | 100 |
| SRP72 | 100 | 100 |
| NFKB1 | 100 | 99.9 |
| MANBA | 100 | 100 |
| TET2 | 100 | 100 |
| CFI | 100 | 97.5 |
| IL2 | 99.9 | 99.9 |
| IL21 | 100 | 100 |
| FAT4 | 100 | 100 |
| LRBA | 100 | 99.2 |
| AGA | 100 | 100 |
| TLR3 | 100 | 100 |
| TERT | 99.8 | 99.9 |
| OTULIN | 92.2 | 96.3 |
| DNAJC21 | 99.7 | 99.9 |
| IL7R | 100 | 100 |
| C9 | 100 | 100 |
| C7 | 100 | 100 |
| C6 | 100 | 100 |
| IL6ST | 99.9 | 99.9 |
| PIK3R1 | 100 | 100 |
| AP3B1 | 99.9 | 97.2 |
| DHFR | 100 | 98.5 |
| TTC37 | 100 | 100 |
| TMEM173 | 100 | 100 |
| SPINK5 | 100 | 99.4 |
| HAVCR2 | 100 | 100 |
| ITK | 100 | 100 |
| IL12B | 100 | 100 |
| DOCK2 | 100 | 100 |
| F12 | 100 | 100 |
| NHP2 | 100 | 100 |
| IRF4 | 100 | 99.2 |
| RIPK1 | 99.9 | 99.9 |
| CFB | 100 | 100 |
| C2 | 100 | 100 |
| SKIV2L | 100 | 100 |
| TAP2 | 100 | 99.5 |
| PSMB8 | 100 | 100 |
| TAP1 | 100 | 100 |
| PSMB9 | 93.9 | 100 |
| SLC39A7 | 100 | 100 |
| TAPBP | 100 | 100 |
| DEF6 | 99.9 | 99.9 |
| RSPH9 | 100 | 100 |
| IL17F | 100 | 100 |
| PGM3 | 100 | 100 |
| SLC35A1 | 100 | 100 |
| BACH2 | 100 | 100 |
| OSTM1 | 100 | 96 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| ZBTB24 | 100 | 100 |
| TRAF3IP2 | 100 | 100 |
| IFNGR1 | 100 | 98.1 |
| TNFAIP3 | 100 | 100 |
| STX11 | 100 | 100 |
| SERAC1 | 100 | 99.7 |
| GTF2H5 | 100 | 100 |
| PLG | 100 | 100 |
| CARD11 | 100 | 100 |
| ACTB | 100 | 100 |
| CLEC7A | 100 | 100 |
| SNX10 | 100 | 100 |
| IKZF1 | 100 | 100 |
| SBDS | 100 | 100 |
| NCF1 | 73.7 | 60.2 |
| SEMA3E | 100 | 95 |
| SAMD9 | 100 | 100 |
| SAMD9L | 100 | 100 |
| ARPC1B | 100 | 100 |
| CFTR | 100 | 99.9 |
| POT1 | 100 | 100 |
| BLK | 100 | 99.8 |
| EXTL3 | 100 | 100 |
| IKBKB | 100 | 100 |
| PRKDC | 100 | 99 |
| MCM4 | 100 | 99.2 |
| CHD7 | 100 | 99.9 |
| CA2 | 100 | 100 |
| NBN | 100 | 100 |
| VPS13B | 100 | 100 |
| GRHL2 | 100 | 100 |
| ANGPT1 | 100 | 100 |
| DGAT1 | 94 | 93 |
| SLC39A4 | 100 | 100 |
| RECQL4 | 100 | 100 |
| DOCK8 | 100 | 100 |
| JAK2 | 99.9 | 98.7 |
| CDKN2B | 100 | 100 |
| DDX58 | 100 | 99.8 |
| RMRP | 100 | 0 |
| PAX5 | 100 | 100 |
| ERCC6L2 | 100 | 100 |
| TLR4 | 100 | 100 |
| C5 | 100 | 99.3 |
| LRRC8A | 100 | 100 |
| CARD9 | 100 | 99.6 |
| C8G | 100 | 99.9 |
| IL2RA | 100 | 100 |
| DCLRE1C | 100 | 99.7 |
| MBL2 | 100 | 100 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| PRF1 | 100 | 100 |
| SLC29A3 | 99.7 | 100 |
| FAS | 100 | 100 |
| HELLS | 99.9 | 98.6 |
| BLNK | 100 | 95.2 |
| NFKB2 | 100 | 100 |
| OBFC1 | 100 | 100 |
| IRF7 | 100 | 99.9 |
| CD81 | 92.8 | 100 |
| STIM1 | 100 | 100 |
| TRIM22 | 100 | 100 |
| CD59 | 100 | 100 |
| RAG1 | 100 | 100 |
| RAG2 | 100 | 100 |
| SLC35C1 | 100 | 100 |
| SERPING1 | 100 | 100 |
| MS4A1 | 100 | 99.4 |
| FERMT3 | 100 | 100 |
| RASGRP2 | 100 | 100 |
| LTBP3 | 100 | 99.5 |
| RNASEH2C | 100 | 100 |
| UNC93B1 | 94.5 | 66.2 |
| TCIRG1 | 100 | 100 |
| FADD | 100 | 100 |
| IL18BP | 100 | 100 |
| CLPB | 100 | 100 |
| CTSC | 100 | 100 |
| MRE11A | 99.9 | 100 |
| ATM | 100 | 99.7 |
| IL10RA | 100 | 100 |
| CD3E | 100 | 100 |
| CD3D | 100 | 100 |
| CD3G | 100 | 100 |
| SLC37A4 | 100 | 100 |
| HYOU1 | 100 | 100 |
| TIRAP | 100 | 100 |
| TNFRSF1A | 100 | 100 |
| CD27 | 100 | 100 |
| C1S | 100 | 100 |
| C1R | 100 | 100 |
| CLEC4D | 100 | 100 |
| AICDA | 100 | 100 |
| KRAS | 100 | 100 |
| EPG5 | 100 | 100 |
| MALT1 | 97.8 | 94.9 |
| CCBE1 | 100 | 100 |
| TNFRSF11A | 94.9 | 94.9 |
| IRAK4 | 100 | 100 |
| KMT2D | 100 | 100 |
| NCKAP1L | 100 | 100 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| STAT2 | 100 | 100 |
| STAT6 | 100 | 100 |
| UNG | 100 | 100 |
| MVK | 100 | 100 |
| SH2B3 | 100 | 100 |
| OAS1 | 100 | 100 |
| ORAI1 | 100 | 98.7 |
| POMP | 99.9 | 84.8 |
| RFXAP | 100 | 99.6 |
| TNFSF11 | 99.9 | 100 |
| LACC1 | 100 | 100 |
| RNASEH2B | 99.6 | 100 |
| PCCA | 99.9 | 100 |
| TPP2 | 99.7 | 98 |
| LIG4 | 100 | 100 |
| PNP | 100 | 100 |
| TRAC | 100 | 48.1 |
| SLC7A7 | 100 | 100 |
| CEBPE | 100 | 100 |
| RNF31 | 100 | 100 |
| IRF9 | 100 | 100 |
| TINF2 | 100 | 100 |
| SRP54 | 100 | 97.4 |
| NFKBIA | 100 | 100 |
| POLE2 | 99.9 | 96.1 |
| SOCS4 | 100 | 100 |
| PSMA3 | 99.9 | 100 |
| MTHFD1 | 100 | 100 |
| BCL11B | 97.6 | 99.8 |
| TRAF3 | 100 | 100 |
| IGHM | 100 | 100 |
| NDNL2 | 100 | 99.9 |
| NOP10 | 100 | 100 |
| RASGRP1 | 100 | 100 |
| INO80 | 100 | 99.9 |
| B2M | 100 | 100 |
| BLOC1S6 | 100 | 99.2 |
| SPPL2A | 94.4 | 96.2 |
| RAB27A | 100 | 100 |
| PSTPIP1 | 100 | 100 |
| CIB1 | 98.9 | 100 |
| BLM | 100 | 99.8 |
| CLCN7 | 100 | 100 |
| MEFV | 100 | 100 |
| DNASE1 | 100 | 100 |
| CREBBP | 100 | 99.4 |
| PMM2 | 100 | 100 |
| CIITA | 100 | 99.9 |
| PARN | 100 | 100 |
| IL21R | 100 | 100 |

| Gennaam | CreV4 (15X) | SSv7 (15X) |
|---|---|---|
| CD19 | 100 | 100 |
| LAT | 100 | 100 |
| CORO1A | 91.9 | 91.9 |
| NOD2 | 100 | 100 |
| USB1 | 100 | 100 |
| RLTPR | 96.1 | 96.5 |
| ACD | 100 | 100 |
| NFAT5 | 99.8 | 99.9 |
| PLCG2 | 100 | 100 |
| IRF8 | 100 | 100 |
| CYBA | 90.9 | 95.5 |
| NLRP1 | 100 | 100 |
| TNFSF12 | 99.5 | 96.2 |
| WRAP53 | 100 | 100 |
| CTC1 | 100 | 100 |
| TNFRSF13B | 100 | 100 |
| SLC46A1 | 100 | 100 |
| FOXN1 | 100 | 100 |
| STAT5B | 100 | 100 |
| STAT3 | 100 | 100 |
| PTRF | 100 | 100 |
| G6PC | 100 | 100 |
| G6PC3 | 100 | 98.1 |
| MAP3K14 | 100 | 100 |
| PLEKHM1 | 100 | 98.8 |
| SMARCD2 | 87.9 | 87.9 |
| CD79B | 100 | 100 |
| UNC13D | 100 | 100 |
| TMC6 | 100 | 100 |
| TMC8 | 100 | 99.5 |
| CARD14 | 99.9 | 100 |
| C17orf62 | 100 | 100 |
| LPIN2 | 100 | 100 |
| PSMG2 | 100 | 100 |
| MC2R | 100 | 100 |