

GENPANEL: Primär Immunbrist (PID)

VERSION: 23

ANTAL GENE: 510

ACD
ACP5
ACTB
ADA
ADAM17
ADAR
AICDA
AIRE
AK2
ALG1
ALPI
ALPK1
AP3B1
AP3D1
APOL1
ARHGEF1
ARPC1B
ARPC5
ATAD3A
ATG4A
ATM
ATP6AP1
B2M
BACH2
BCL10
BCL11B
BLNK
BTK
C16orf57 (USB1)
C1QA
C1QB
C1QBP
C1QC
C1R
C1S
C2
C2orf69
C3
C4A
C4B
C5
C6
C7

C8A
C8B
C8G
C9
CA2
CARD11/CARMA11/BIMP3
CARD14 (CARMA2)
CARD8
CARD9
CARMIL2
CASP10
CASP8
CBL
CCR2
CD19
CD21 / CR2
CD247
CD27 / TNFRSF7
CD274
CD28
CD3D
CD3E
CD3G
CD4
CD40
CD40LG
CD46
CD48
CD55
CD59
CD70
CD79A
CD79B
CD81
CD8A
CDC42
CDCA7
CEBPE
CECR1 / ADA2
CFB
CFD
CFHR1
CFHR3
CFI
CHD7
CHS1 / LYST

CHUK
CIAS1 / PYPAF1 /NLRP3
CIB1
CLCN7
CLEC16A
CLEC7A
CLPB
COH1 / VPS13B
COL7A1
COLEC11/CLK1
COPA
COPG1
CORO1A
CRACR2A
CSF2RB
CSF3R
CTC1
CTLA4
CTNNB1
CTPS1
CTSC (Cathepsin C)
CWF19L2
CXCR2
CXCR4
CYBA / p22-PHOX
CYBB / p91-PHOX
DBR1
DCLRE1B
DCLRE1C
DDX58 /RIGI
DEF6
DIAPH1
DKC1
DNAJC21
DNASE1L3
DNASE2
DNMT3B
DOCK11
DOCK2
DOCK8
DUOX1
DUOX2
DUOXA1
EFL1
ELANE
ELF4

EPG5
ERBIN / ERBB2IP
ERCC6L2
EROS/CYBC1
EXTL3
EZR
F12
FAAP24
FADD
FAS (TNFRSF6/CD95)
FASLG (TNFSF6 /CD178)
FCGR3A/CD16B
FCHO1
FCN3
FERMT1
FLT3LG
FNIP1
FOXI3
FOXN1
FOXP3
FPR1
G6PC (formerly G6PT)
G6PC3
G6PD
GATA1
GATA2
GFI1 / ZNF163
GIMAP5
GIMAP6
GINS1
GINS4
GMCSFR / CSF2RA
GNAI2
GTF3A
GUCY2C
HAVCR2 (TIM3)
HAX1
HCK
HELLS
HF1 / CFH
HMOX1
HOIP / RNF31
HPS1
HPS4
HPS6
HYOU1

ICOS
ICOSLG
IFIH1 (GOF)
IFNAR1
IFNAR2
IFNG
IFNGR1
IFNGR2
IGHM
IGLL1 (λ 5)
IKBKB
IKBKE
IKBKG
IKZF1 (Ikaros)
IKZF2
IKZF3
IL10
IL10RA
IL10RB
IL12B/23 (p40)
IL12RB1
IL12RB2
IL17F
IL17RA
IL17RC
IL18BP
IL1RN
IL21
IL21R
IL23R
IL27RA
IL2RA
IL2RB
IL2RG
IL36RN
IL6R
IL6ST
IL7
IL7R
INO80
IRAK1
IRAK4
IRF1
IRF2BP2
IRF3
IRF4

IRF7
IRF8
IRF9
ISG15
ITCH
ITGB2 (CD18)
ITK
ITPKB
JAGN1
JAK1
JAK3
KIND3 / KINDLIN3 / FERMT3
KMT2D
KRAS
LACC1
LAT
LCK
LCP1
LCP2
LIG1
LIG4
LPIN2
LRBA
LRRC8A
LSM11
LTBR
MADD
MAGT1
MALT1
MAN2B2
MAP1LC3B2
MAP3K14
MAP4K1
MAPK8
MASP1
MASP2
MBL2
MCM10
MCM4
MEFV
MHC2TA / CIITA
MKL1
MOGS
MRE11A
MS4A1 (CD20)
MSH6

MSN
MTHFD1
MVK
MYD88
MYO5A
MYSM1
NAF1
NALP12 /NLRP12
NBAS
NBEAL2
NBS1 / NBN
NCF1 / p47-PHOX
NCF2 /p67-PHOX
NCF4 / p40-PHOX
NCKAP1L
NEIL3
NFAT5
NFE2L2
NFKB1
NFKB2
NFKBIA
NFKBID
NHEJ1
NHP2
NKX2-5
NLRC4
NLRP1
NOD2
NOLA3 / NOP10
NOS2
NPC1
NRAS
NSMCE3
NUDCD3
OAS1
OAS2
ORAI1
OSTM1
OTULIN
OX40 / TNFRSF4
P14 / MAPBPIP / LAMTOR2
PARN
PAX1
PDCD1
PDLIM1
PEPD

PFC / CFP
PGM3
PIGA
PIK3CA
PIK3CD
PIK3CG
PIK3R1
PLCG1
PLCG2
PLDN / BLOC1S6
PLEKHM1
PLVAP
PMS2
PNP
POLA1
POLD1
POLD2
POLD3
POLE
POLE2
POLR3A
POLR3C
POLR3E
POLR3F
POMP
POT1
POU2AF1
PRF1 (Perforin)
PRKCD
PRKDC
PSMA3
PSMB10
PSMB4
PSMB8
PSMB9
PSMG2
PSTPIP1
PTCRA
PTEN
PTPN2
PTPRC
RAB27A
RAC2
RAG1
RAG2
RANBP2

RAP1B
RASGRP1
RBCK1 / HOIL1 / XAP3
RC3H1
RECQL3 / BLM
RECQL4
REL
RELA
RELB
RFX5
RFXANK
RFXAP
RHOG
RHOH
RIPK1
RIPK3
RMRP
RNASEH2A
RNASEH2B
RNASEH2C
RNASEL
RNF168
RNU4ATAC (MOPD1)
RNU7-1
RORC
RPA1
RPSA
RTEL1
SAMD9
SAMD9L
SAMHD1
SASH3
SBDS
SEC61A1
SEMA3E
SERPING1 / C1INH
SH2D1A
SH3BP2
SH3KBP1
SHARPIN
SKIV2L
SLC11A1 / NRAMP1
SLC26A3
SLC29A3
SLC35C1 / FUT1 (Fucose transp.)
SLC37A4

SLC46A1
SLC7A7
SLC9A3
SLCO2A1
SMARCAL1
SMARCD2
SNORA31
SNX10
SOCS1
SP110
SPI1
SPINK5
SPPL2A
SRP54
STAT1
STAT2
STAT3
STAT4
STAT5B
STAT6
STIM1
STK4
STN1
STX11
STXBP2
STXBP3
SYK
TAP1
TAP2
TAPBP (Tapasin)
TAZ
TBK1
TBX1
TBX21
TCF3 / E2A
TCIRG1
TCN2
TERC
TERT
TET2
TFRC
TGFB1
TGFB1R1
TGFB1R2
THBD
TICAM1 / TRIF

TINF2
TIRAP
TLR2
TLR3
TLR7
TLR8
TMC6 / EVER1
TMC8 / EVER2
TMEFF1
TMEM173
TNF
TNFAIP3
TNFRSF11A
TNFRSF13B (TACI)
TNFRSF13C (BAFF-R)
TNFRSF1A
TNFRSF9
TNFSF10 (TRAIL)
TNFSF11
TNFSF13 (APRIL)
TOP2B
TP63
TPP2
TRAC
TRAF3
TRAF3IP2 / ACT1
TREX1
TRIM22
TRNT1
TTC37
TTC7A
TWEAK / TNFSF12
TYK2
TYMS
UBA1
UNC13D
UNC93B1
UNG
USP18
USP43
VAV1
VPS45
WAS
WDR1
WIPF1
WRAP53

XIAP

ZAP70

ZBTB24 (ICF2)

ZCCHC8

ZIP7 / SLC39A7

ZNF341

ZNFX1