

Indicazione: Difetto immunitario primitivo	
Gene	Regione non coperte per disegno del pannello
ACD	
ACP5	
ACTB	
ADA	
ADAM17	
ADAR	
AICDA	
AIRE	
AK2	
ALPI	
AP1S3	
AP3B1	
AP3D1	
APOL1	
ARHGEF1	
ARPC1B	
ATAD3A	
ATG4A	
ATM	
ATP6AP1	
ATP6AP2	
B2M	
BACH2	
BCL10	
BCL11B	
BLM	
BLNK	
BPIFA1	
BTK	
C1QA	
C1QB	
C1QC	
C1R	
C1S	
C2	
C2ORF69	
C3	
C4A	parzialmente coperto con qualità bassa
C4B	parzialmente coperto con qualità bassa
C5	
C6	
C7	
C8A	
C8B	
C8G	
C9	
CARD11	
CARD14	
CARD9	
CARMIL2	
CASP10	
CASP8	
CBLB	chr3:105868164-105868243 NM_001321786 NM_001321789.1 chr3:105724021-105724045 NM_001321820
CCBE1	
CD19	
CD247	
CD27	
CD3D	
CD3E	
CD3G	
CD40	
CD40LG	

CD46	
CD55	chr1:207340386-207340649 NM_001114752 NM_001300902 chr1:207347442-207347566 NM_001300903
CD59	
CD70	
CD79A	
CD79B	
CD81	
CD8A	
CDC42	
CDCA7	
CEBPE	
CECR1	
CFB	
CFD	
CFH	
CFI	chr4:109734654-109734801 NM_001375278 NM_001375279 NM_001375280
CFP	
CHD7	
CHUK	
CIB1	
CIITA	
CLPB	
COPA	
COPG1	
CORO1A	
CR2	
CRACR2A	
CSF2RB	
CSF3R	
CTC1	
CTLA4	
CTNBL1	
CTPS1	
CTSC	
CXCR2	
CXCR4	
CYBA	
CYBB	
CYBC1	
DBF4	
DBR1	
DCLRE1C	chr10:14899245-14899317 NM_001350965 NM_001350966 NM_001350967
DEF6	
DKC1	
DNAJC21	
DNASE1	
DNASE1L3	
DNASE2	
DNMT3B	
DOCK11	
DOCK2	
DOCK8	
DPP9	
EFL1	
ELANE	
ELF4	
EPG5	
ERBIN	
ERCC6L2	
ETS1	
EXTL3	
FAAP24	
FADD	
FAS	

FASLG	
FAT4	
FCGR3A	
FCHO1	
FCN3	
FERMT1	
FERMT3	
FGL2	
FNIP1	
FOXI3	esone 1 parzialmente coperto
FOXN1	
FOXP3	
FPR1	
G6PC3	
G6PD	
GATA2	
GFI1	
GIMAP5	
GINS1	
GJA1	
HAVCR2	
HAX1	
HCK	
HELLS	
HMOX1	
HYOU1	
ICOS	
ICOSLG	parzialmente coperto con qualità bassa
IFIH1	
IFNAR1	chr21:33357736-33357761 NM_001384498
IFNAR2	chr21:33264929-33264973 NM_001385055
IFNG	
IFNGR1	
IFNGR2	
IFNL2	
IFNL3	
IGHM	
IGKC	
IGLL1	
IKBKB	
IKZF1	
IKZF3	
IL10	
IL10RA	
IL10RB	
IL12B	
IL12RB1	chr19:18071204-18071338 NM_153701
IL12RB2	
IL17F	
IL17RA	
IL17RC	
IL18BP	
IL1RN	
IL21	
IL21R	
IL23R	
IL2RA	
IL2RB	
IL2RG	
IL33	
IL36RN	
IL6R	chr1:154437430-154437549 NM_001206866
IL6ST	
IL7R	

INO80	
IRAK1	
IRAK4	
IRF2BP2	
IRF3	
IRF4	
IRF7	
IRF8	
IRF9	
ISG15	
ITCH	chr20:34417112-34417244 NM_001257137 NM_001324197
ITGB2	
ITK	
ITPKB	
ITPR3	
JAGN1	
JAK1	
JAK3	
KDM6A	
KMT2A	
KMT2D	
KRAS	
LACC1	
LAMTOR2	
LAT	
LCK	
LCP2	
LIG1	
LIG4	
LPIN2	
LRBA	
LRRC32	
LSM11	
LY96	
LYST	
MAGT1	
MALT1	
MAN2B2	
MAP1LC3B2	
MAP3K14	
MAPK8	
MASP2	
MCM10	
MCM4	
MEFV	
MOGS	
MRTFA	
MS4A1	
MSH6	
MSN	
MTHFD1	
MVK	
MYD88	
MYSM1	
NBAS	
NBN	
NCF1	parzialmente coperto con qualità bassa
NCF2	
NCF4	
NCKAP1L	
NCSTN	
NEIL3	
NFAT5	
NFE2L2	

NFKB1	
NFKB2	
NFKBIA	
NHEJ1	
NHP2	
NLRC4	
NLRP1	
NLRP12	
NLRP3	
NOD2	
NOP10	
NOS2	
NRAS	
NSMCE3	
OAS1	
OAS2	
ORAI1	
OTULIN	
PARN	
PAX1	
PDCD1	
PEPD	
PGM3	
PI4KA	
PIK3CD	
PIK3CG	
PIK3R1	
PLCG2	
PNP	
POLA1	
POLD1	
POLD2	
POLE	
POLE2	
POLR3A	
POLR3C	
POLR3F	
POMP	
POT1	
POU2AF1	
PRF1	
PRKCD	
PSEN1	
PSENFEN	
PSMA3	
PSMB10	
PSMB4	
PSMB8	
PSMB9	
PSMG2	
PSTPIP1	
PTEN	
PTPN2	
PTPRC	
RAB27A	
RAC2	
RAG1	
RAG2	
RANBP2	
RAP1A	
RAP1B	
RASGRP1	
RBCK1	
RC3H1	

REL	
RELA	
RELB	
RFWD3	
RFX5	
RFXANK	chr19:19200814-19200898 NM_001370237 NM_001370238
RFXAP	
RHBDF2	
RHOG	
RHOH	
RIPK1	
RMRP	
RNASEH2A	
RNASEH2B	
RNASEH2C	
RNF168	
RNF31	
RNU4ATAC	
RNU7-1	
RORC	
RPA1	
RPSA	
RTKL1	
SAMD9	
SAMD9L	
SAMHD1	
SASH3	
SBDS	
SEC61A1	
SEMA3E	
SERPING1	
SH2D1A	
SH3BP2	esone 1 parzialmente coperto NM_001145856
SH3KBP1	
SHARPIN	
SKIV2L	
SLC29A3	
SLC35C1	
SLC37A4	
SLC39A7	
SLC46A1	
SLC7A7	
SMARCAL1	
SMARCD2	
SNM1B/DCLRE1B	
SNORA31	
SOCS1	
SP110	
SPI1	
SPINK5	
SPPL2A	
SRP19	
SRP54	
SRP72	
SRPRA	
STAT1	
STAT2	
STAT3	
STAT5B	
STAT6	
STIM1	
STK4	
STN1	
STX11	

STXBP2	
SYK	
TAP1	
TAP2	
TAPBP	
TAZ	
TBK1	
TBX1	chr22:19760839-19761106 NM_005992 NM_080646 NM_080647 NM_001379200
TBX2	chr17:61400333-61400433 NM_005994
TBX21	
TCF3	
TCN2	
TERC	
TERT	
TET2	
TFRC	
TGFB1	
TGFBR1	chr9:99105201-99105307 NM_001130916 NM_001306210 NM_004612
TGFBR2	
THBD	
TICAM1	
TINF2	
TIRAP	
TLR3	
TLR4	
TLR7	
TLR8	
TMC6	
TMC8	
TMEM173/STING1	
TNFAIP3	
TNFRSF11A	chr18:62325348-62325373 NM_001270949 NM_001270950 NM_001270951 NM_001278268 NM_003839
TNFRSF13B	
TNFRSF13C	
TNFRSF17	
TNFRSF1A	chr12:6331871-6331972 NM_001346092
TNFRSF4	
TNFRSF9	
TNFSF11	
TNFSF12	
TNFSF13	
TOM1	
TOP1MT	
TOP2B	
TP53	
TPP2	
TRAC	
TRAF3	
TRAF3IP2	
TREX1	
TRIM22	
TRNT1	
TTC37	
TTC7A	chr2:46917191-46917282 NM_001288953
TYK2	chr19:10354797-10354887 NM_001385203
UBA1	
UNC13D	
UNC93B1	
UNG	
USB1	
USP18	esone 11 parzialmente coperto
VPS13B	
VPS45	chr1:150142833-150142936 NM_001279353
WAS	esone 10 parzialmente coperto

WDR1	
WIPF1	
WRAP53	
XIAP	
ZAP70	
ZBTB24	
ZIC3	
ZNF341	
ZNFX1	

Indicazione: Piastrinopenia congenita	
Gene	Regione non coperte per disegno del pannello
ABCG5	
ABCG8	
ACTN1	
ADAMTS13	
ANKRD26	
ANO6	
CDC42	
CYCS	
DIAPH1	
EPHB2	chr1:22710978-22711048 NM_001309192 NM_001309193 NM_004442 NM_017449
ETV6	
FLI1	
FLNA	
FYB1	
GALE	
GATA1	
GFI1B	
GNE	
GP1BA	
GP1BB	
GP6	
GP9	
HOXA11	
IKZF5	
ITGA2B	
ITGB3	
KAT6A	
KDSR	
LCP2	
MASTL	
MECOM	
MPIG6B	
MPL	
MYH9	
NBEAL2	
P2RY12	
PLA2G4A	
PLAU	
PRKACG	
PTPRJ	
RASGRP2	
RBM8A	
RUNX1	
SLC35A1	
SLFN14	
SRC	
TBXA2R	
THPO	
TPM4	
TRPM7	
TUBB1	
VWF	esone 26 parzialmente coperto
WAS	esone 10 parzialmente coperto

Indicazione: Anemia ereditaria	
Gene	Regione non coperte per disegno del pannello
ABCB6	
ABCB7	
ABCG5	
ABCG8	
ADK	
AK1	
ALAS2	
ALDOA	
ANK1	
ATP11C	chrX:139732431-139732530 NM_001353811
BCL11A	
BPGM	
C15orf41/CDIN1	
CAD	
CDAN1	
COX4I2	
CYB5R3	
DKC1	
ENO1	
EPB41	
EPB42	
EPO	
G6PD	
GATA1	
GATA2	
GBA	
GCLC	
GCLM	
GPI	
GPX1	
GSR	
GSS	
HBB	
HFE	
HK1	chr10:69292253-69292355 NM_001322365
HSCB	
HSPA9	
KCNN4	
KIF23	
KLF1	
LARS2	
LCP2	
LPIN2	
NDUFB11	
NHP2	
NOP10	
NT5C3A	
PFKL	
PFKM	
PGK1	
PGM1	
PIEZO1	
PKLR	
PUS1	

RAD51	chr15:40701801-40701931 NM_133487 NM_001164269
RHAG	
RPL11	
RPL15	chr3:23921593-23921680 NM_001253384
RPL18	
RPL19	
RPL23	
RPL26	
RPL27	
RPL31	
RPL35	
RPL35A	
RPL36	
RPL5	
RPL9	
RPS10	
RPS14	
RPS15	
RPS15A	
RPS17	
RPS19	
RPS24	
RPS26	
RPS27	
RPS27A	
RPS28	
RPS29	
RPS7	
SBDS	
SEC23B	
SLC11A2	
SLC19A1	
SLC19A2	
SLC25A38	
SLC2A1	
SLC46A1	
SLC4A1	
SPTA1	
SPTB	
STEAP3	
TERC	
TERT	
TINF2	
TMPRSS6	
TPI1	
TSR2	
UBE2T	
XRCC4	
YARS2	

Indicazione: Insufficienza midollare ereditaria	
Gene	Regione non coperte per disegno del pannello
ABCB7	
ABCD4	
ABL1	
ACD	
ADA	
ADA2	
AIRE	
AK2	
ALAS2	
ANKRD26	
ATM	
ATR	
BRCA1	chr17:43079329-43079404 NM_007300
BRCA2	
BRIP1	
C15orf41	
CBL	
CD27	
CDAN1	
CLCN7	
CLPB	
CTC1	
CXCR2	
CXCR4	
DBF4	
DDX41	
DHFR	
DKC1	
DNAJC21	
DUT	
EFL1	
EIF2AK3	
ELANE	
EPO	
ERCC4	
ERCC6L2	
ETV6	
EXOC3L2	
FANCA	
FANCB	
FANCC	
FANCD2	
FANCE	
FANCF	
FANCG	
FANCI	
FANCL	
FANCM	
G6PC3	
GATA1	
GATA2	
GFI1B	
GLRX5	
HAX1	

HOXA11
IVD
KAT6A
KIF23
KLF1
LIG4
MAD2L2
MBD4
MDM4
MECOM
MPL
NAF1
NBEAL2
NHP2
NOP10
PALB2
PARN
POT1
PSTPIP1
PUS1
RAD51
RAD51C
RBM8A
RMRP
RPA1
RPL11
RPL15
RPL19
RPL23
RPL26
RPL27
RPL31
RPL35A
RPL36
RPL5
RPL9
RPS10
RPS14
RPS15
RPS17
RPS19
RPS24
RPS26
RPS27
RPS27A
RPS28
RPS29
RPS7
RTEL1
RUNX1
SAMD9
SAMD9L
SBDS
SEC23B
SLC19A2
SLC25A38

SLC37A4	
SLC46A1	
SLX4	
SMARCAL1	
SNM1B/DCLRE1B	
SP1	
SRP19	
SRP54	
SRP72	
SRPRA	
TAZ	
TBXAS1	
TCIRG1	
TERC	
TERT	
THPO	
TINF2	
TP53	
TSR2	
UBE2T	
USB1	
WAS	esone 10 parzialmente coperto
WRAP53	
XRCC2	
XRCC4	
ZCCHC8	

Indicazione: Predisposizione genetica a neoplasia	
Gene	Regione non coperte per disegno del pannello
ALK	
ANKRD26	
APC	
ASXL1	chr20:32372159-32372171 NM_001164603
ATG2B	
ATM	
ATRX	
BAP1	
BARD1	
BLM	
BMPR1A	
BRCA1	chr17:43079329-43079404 NM_007300
BRCA2	
BRIP1	
BUB1B	
CARD11	
CARMIL2	
CASP10	
CD27	
CDC73	
CDH1	
CDK4	
CDKN1B	
CDKN1C	esone 1 parzialmente coperto
CDKN2A	
CEBPA	
CHEK2	
CREBBP	
CTC1	
CTR9	
DDB2	
DDX41	
DHX34	
DICER1	
DIS3L2	
DKC1	
DOCK8	
ELANE	
EP300	
EPCAM	
ERCC1	
ERCC2	
ERCC3	
ERCC4	
ERCC5	
ETV6	
EXT1	
EXT2	
EZH2	
FAH	
FANCA	
FANCB	
FANCC	
FANCD2	

FANCE
FANCF
FANCG
FANCI
FANCL
FANCM
FAS
FASLG
FBXW7
FH
FLCN
GALNT14
GATA1
GATA2
GPC3
GPC4
GRHL2
GSKIP
HAX1
HCLS1
HLTF
HOXB13
HRAS
IGF2
IKZF1
ITK
JAK2
KDM3B
KIT
KLHDC8B
L2HGDH
LAPTM5
LZTR1
MAGT1
MAP2K1
MAP2K2
MAX
MEN1
MITF
MLH1
MSH2
MSH6
MUTYH
NAF1
NBN
NF1
NF2
NHP2
NKX2-1
NOP10
NPAT
NR3C1
NSD1
NT5C2
NTRK1
NYNRIN

PALB2	
PARN	
PAX5	
PDGFRA	chr4:64240048-54240120 NM_001347828
PHF6	
PHOX2B	
PIK3CA	
PIK3CD	
PIK3R1	
PMS2	
POLH	
PPM1D	
PRKAR1A	
PTCH1	
PTCH2	
PTEN	
PTPN11	
RAD51	chr15:40701801-40701931 NM_133487 NM_001164269
RAD51B	
RAD51C	
RAD51D	
RAF1	
RB1	
RECQL4	
REST	
RET	
RHBDF2	
RIT1	
RNASEL	
RPL11	
RPL15	
RPL19	
RPL23	
RPL26	
RPL27	
RPL31	
RPL35A	
RPL36	
RPL5	
RPL9	
RPS10	
RPS14	
RPS15	
RPS17	
RPS19	
RPS24	
RPS26	
RPS27	
RPS27A	
RPS28	
RPS29	
RPS7	
RTEL1	
RUNX1	
SAMD9	
SAMD9L	

SBDS	
SDHA	
SDHAF2	
SDHB	
SDHC	
SDHD	chr11:112093075-112093202 NM_001276506
SETBP1	
SH2B3	
SH2D1A	
SHOC2	
SLX4	
SMAD4	
SMARCA4	
SMARCB1	
SMARCE1	
SMO	
SOS1	
SOS2	
SPRED1	
SRP72	
STAT3	
STK11	
STN1	
SUFU	
TERC	
TERT	
TINF2	
TMEM127	
TP53	
TRIM28	
TRIM37	
TRIP13	
TSC1	
TSC2	
TSR2	
UBE2T	
USB1	
VHL	
WAS	
WRAP53	
WRN	
WT1	
WWP1	
XPA	
XPC	
XRCC2	