

Indicazione: Difetto immunitario primitivo	
Gene	Regione non coperte per disegno del pannello
ACD	
ACP5	
ACTB	
ADA	
CECR1/ADA2	
ADAM17	
ADAR	
AICDA	
AIRE	
AK2	
ALPI	
AP1S3	chr2:223752826-223752889
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	
CCBE1	

CD19	
CD247	
CD27	
CD3D	
CD3E	
CD3G	
CD40	
CD40LG	
CD46	
CD55	esone 10 non coperto (chr1:207340386-207340649 NM_001300903, NM_001300902, NM_001114752)
CD59	
CD70	
CD79A	
CD79B	
CD81	
CD8A	
CDC42	
CDCA7	
CEBPE	
CFB	
CFD	
CFH	
CFI	
CFP	
CHD7	
CIB1	
CIITA	
CLPB	
COPA	
CORO1A	esone 10 parzialmente coperto con reads a bassa qualità; esone 11 bassa qualità
CR2	
CSF2RA	bassa qualità
CSF2RB	
CSF3R	
CTC1	
CTLA4	
CTNBL1	
CTPS1	
CTSC	
CXCR2	
CXCR4	
CYBA	
CYBB	
CYBC1	
DBF4	
DBR1	
DCLRE1C	
DEF6	
DKC1	
DNAJC21	
DNASE1	
DNASE1L3	
DNASE2	
DNMT3B	
DOCK2	
DOCK8	

EFL1	
ELANE	
EPG5	
ERBIN	
ERCC6L2	
ETS1	
EXTL3	
FAAP24	
FADD	
FAS	
FASLG	
FAT4	
FCGR3A	
FCHO1	
FCN3	
FERMT1	
FERMT3	
FNIP1	
FOXI3	
FOXN1	
FOXP3	
FPR1	
G6PC3	
G6PD	
GATA2	
GFI1	
GIMAP5	
GINS1	
GJA1	
HAVCR2	
HAX1	
HCK	
HELLS	
HMOX1	
HYOU1	
ICOS	
ICOSLG	bassa qualità
IFIH1	
IFNAR1	esone 12 non coperto (chr21:33357736-33357761 NM_001384498)
IFNAR2	esone 10 non coperto (chr21:33264929-33264973 NM_001385055)
IFNG	
IFNGR1	
IFNGR2	
IFNL2	
IFNL3	
IGHM	
IGKC	
IGLL1	
IKBKB	
IKBKG	bassa qualità
IKZF1	
IKZF3	
IL10	
IL10RA	
IL10RB	
IL12B	

IL12RB1	esone 10 (chr19:18071204-18071338 NM_153701)
IL12RB2	
IL17F	
IL17RA	
IL17RC	
IL18BP	
IL1RN	
IL21	
IL21R	
IL23R	
IL2RA	
IL2RB	
IL2RG	
IL33	
IL36RN	
IL6R	esone 7 non coperto (chr1:154437430-154437549 NM_001206866)
IL6ST	
IL7R	
INO80	
IRAK1	
IRAK4	
IRF2BP2	
IRF3	
IRF4	
IRF7	
IRF8	
IRF9	
ISG15	
ITCH	esone 7 non coperto (chr20:34417112-34417244 NM_001257137, NM_001324197)
ITGB2	
ITK	
ITPR3	
JAGN1	
JAK1	
JAK3	
KDM6A	
KMT2A	
KMT2D	
KRAS	
LACC1	
LAMTOR2	
LAT	
LCK	
LCP2	
LIG1	
LIG4	
LPIN2	
LRBA	
LRRC32	
LSM11	
LYST	
MAGT1	regione non coperta (chrX:77868637-77868649)
MALT1	
MAN2B2	
MAP1LC3B2	
MAP3K14	

MAPK8	
MASP2	
MCM10	
MCM4	regione non coperta (chr8:47969278-47969401)
MEFV	regione non coperta (chr16:3251956-3252049)
MOGS	
MRTFA	
MS4A1	
MSH6	
MSN	
MTHFD1	
MVK	
MYD88	
MYSM1	
NBAS	
NBN	
NCF1	bassa qualità
NCF2	
NCF4	
NCKAP1L	
NCSTN	
NEIL3	
NFAT5	
NFE2L2	regione non coperta (chr2:177227908-177227953)
NFKB1	
NFKB2	
NFKBIA	
NHEJ1	
NHP2	
NLRC4	
NLRP1	
NLRP12	
NLRP3	
NOD2	
NOP10	
NOS2	
NRAS	
NSMCE3	
OAS1	
ORAI1	
OTULIN	
PARN	regione non coperta (chr16:14482288-14482388)
PAX1	
PDCD1	
PEPD	
PGM3	
PI4KA	
PIK3CD	
PIK3CG	
PIK3R1	
PLCG2	
PNP	
POLA1	
POLD1	
POLD2	
POLE	

POLE2	
POLR3A	
POLR3C	
POLR3F	
POMP	
POT1	
POU2AF1	
PRF1	
PRKCD	
PSEN1	
PSENE1	
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	esone 9 non coperto (chr19:19200814-19200898 NM_001370238, NM_001370237)
RFXAP	
RHBDF2	
RHOH	
RIPK1	
RMRP	
RNASEH2A	
RNASEH2B	regione non coperta (chr13:50959473-50959596)
RNASEH2C	
RNF168	
RNF31	
RNU4ATAC	
RNU7-1	
RORC	
RPA1	
RPSA	
RTEL1	
SAMD9	
SAMD9L	
SAMHD1	

SASH3	
SBDS	
SEC61A1	
SEMA3E	
SERPING1	
SH2D1A	
SH3BP2	
SH3KBP1	
SHARPIN	
SKIV2L	
SLC29A3	
SLC35C1	
SLC37A4	
SLC39A7	
SLC46A1	
SLC7A7	
SMARCAL1	
SMARCD2	
DCLRE1B	
SNORA31	
SOCS1	
SP110	
SPINK5	
SPPL2A	
SRP54	
SRP72	
STAT1	
STAT2	
STAT3	
STAT5B	
STAT6	
STIM1	
STK4	
STN1	
STX11	
STXBP2	
SYK	
TAP1	
TAP2	
TAPBP	regione non coperta (chr6:33308104-33308158)
TAZ	
TBK1	
TBX1	
TBX2	
TBX21	
TCF3	
TCN2	
TERC	
TERT	
TET2	
TFRC	
TGFB1	
TGFBR1	
TGFBR2	
THBD	
TICAM1	

TINF2	
TIRAP	
TLR3	
TLR7	
TLR8	
TMC6	
TMC8	
TMEM173	
TNFAIP3	
TNFRSF11A	
TNFRSF13B	
TNFRSF13C	
TNFRSF17	
TNFRSF1A	regione non coperta (chr12:6331875-6331993)
TNFRSF4	
TNFRSF9	
TNFSF11	
TNFSF12	
TNFSF13	
TOM1	
TOP2B	
TP53	
TPP2	
TRAC	
TRAF3	
TRAF3IP2	
TREX1	
TRIM22	
TRNT1	
TTC37	
TTC7A	esone 2 non coperto (chr2:46917191-46917282 NM_001288593)
TYK2	esone 19 non coperto (chr19:10354797-10354887 NM_001385203)
UBA1	
UNC13D	
UNC93B1	
UNG	
USB1	
USP18	
VPS13B	
VPS45	esone 13 non coperto (chr1:150142833-150142936 NM_001279353)
WAS	
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	
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	
WAS	

Indicazione: Anemia ereditaria	
Gene	Regione non coperte per disegno del pannello
ABCB6	
ABCB7	
ABCG5	
ABCG8	
ADK	chr10:74686698-74686820
AK1	
ALAS2	
ALDOA	
ANK1	
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	esone 5 non coperto (chr10:69292253-69292355 NM_001322365)
KCNN4	
KIF23	
KLF1	
LCP2	
LPIN2	
NHP2	
NOP10	
NT5C3A	
PFKL	
PFKM	
PGK1	
PGM1	
PIZO1	
PKLR	
PUS1	
RAD51	esone 4 non coperto (chr15:40701801-40701931 NM_133487, NM_001164269)
RHAG	
RPL11	
RPL15	esone 5 non coperto (chr3:23921593-23921680 NM_001253384)
RPL19	

RPL23	
RPL26	
RPL27	
RPL35A	
RPL36	
RPL5	
RPL9	
RPS10	
RPS14	
RPS15	
RPS17	
RPS19	
RPS24	
RPS26	
RPS27	
RPS27A	
RPS28	
RPS29	
RPS7	
SBDS	
SEC23B	
SLC11A2	
SLC19A1	
SLC19A2	
SLC25A38	
SLC2A1	
SLC46A1	
SLC4A1	
SPTA1	
SPTB	
TERC	
TERT	
TINF2	
TMPRSS6	
TPI1	
TSR2	
UBE2T	
XRCC4	

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	esone 13 non coperto (chr17:43079329-43079404 NM_007300)
BRCA2	
BRIP1	
C15orf41/CDIN1	
CBL	
CD27	
CDAN1	
CLCN7	
CLPB	
CTC1	
CXCR2	
CXCR4	
DBF4	
DDX41	
DHFR	
DKC1	
DNAJC21	
EFL1	
EIF2AK3	
ELANE	
EPO	
ERCC4	
ERCC6L2	
ETV6	
FANCA	
FANCB	
FANCC	
FANCD2	
FANCE	
FANCF	
FANCG	
FANCI	
FANCL	regione non coperta (chr2:58213654-58213706)
FANCM	
G6PC3	
GATA1	
GATA2	
GFI1B	
GLRX5	
HAX1	
HOXA11	
IVD	

KAT6A	
KIF23	
KLF1	
LIG4	
MAD2L2	
MECOM	
MPL	
NAF1	
NBEAL2	
NHP2	
NOP10	
PALB2	
PARN	regione non coperta (chr16:14482288-14482388)
POT1	
PSTPIP1	
PUS1	
RAD51	esone 4 non coperto (chr15:40701801-40701931 NM_133487, NM_001164269)
RAD51C	
RBM8A	
RMRP	
RPA1	
RPL11	
RPL15	esone 5 non coperto (chr3:23921593-23921680 NM_001253384)
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	

DCLRE1B	
SP1	
SRP54	
SRP72	
TAZ	
TBXAS1	
TCIRG1	
TERC	
TERT	
THPO	
TINF2	
TP53	
TSR2	
UBE2T	
USB1	
WAS	
WRAP53	
XRCC2	
XRCC4	

Indicazione: Predisposizione genetica a neoplasia	
Gene	Regione non coperte per disegno del pannello
ALK	
ANKRD26	
APC	
ASXL1	esone 5 non coperto (chr20:32372159-32372171 NM_001164603)
ATG2B	
ATM	
ATRX	
BAP1	
BARD1	
BLM	
BMPR1A	
BRCA1	esone 13 non coperto (chr17:43079329-43079404 NM_007300)
BRCA2	
BRIP1	
BUB1B	
CARD11	
CARMIL2	
CASP10	
CD27	
CDC73	
CDH1	
CDK4	
CDKN1B	
CDKN1C	
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	regione non coperta (chr2:58213654-58213706)
FANCM	
FAS	
FASLG	
FBXW7	
FH	
FLCN	
GALNT14	
GATA1	
GATA2	
GPC3	
GPC4	
GRHL2	
GSKIP	
HAX1	
HCLS1	
HOXB13	
HRAS	
IGF2	
IKZF1	
ITK	
JAK2	
KDM3B	
KIT	
KLHDC8B	
L2HGDH	
LPTM5	
LZTR1	
MAGT1	regione non coperta (chrX:77868637-77868649)
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	regione non coperta (chr16:14482288-14482388)
PAX5	
PDGFRA	esone 2 non coperto (chr4:54240048-54240120 NM_001347828)
PHF6	
PHOX2B	
PIK3CA	
PIK3CD	
PIK3R1	
PMS2	
POLH	
PPM1D	
PRKAR1A	
PTCH1	
PTCH2	
PTEN	
PTPN11	
RAD51	esone 4 non coperto (chr15:40701801-40701931 NM_133487, NM_001164269)
RAD51B	
RAD51C	
RAD51D	
RAF1	
RB1	
RECQL4	
REST	
RET	
RHBDF2	
RIT1	
RNASEL	
RPL11	
RPL15	esone 5 non coperto (chr3:23921593-23921680 NM_001253384)
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	esone 4 non coperto (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	regione non coperta (chr17:59090055-59090092)
TRIP13	
TSC1	
TSC2	
TSR2	
UBE2T	
USB1	
VHL	
WAS	
WRAP53	
WRN	
WT1	
WWP1	
XPA	
XPC	
XRCC2	