Immunsviktsykdommer

Genpanel, versjon v03

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

Gen	Transkript	>10x	Fenotype
ACD	NM_001082487.1	100%	?Dyskeratosis congenita, autosomal recessive 7 OMIM ?Dyskeratosis congenita, autosomal dominant 6 OMIM
ACP5	NM_001111035.1	100%	Spondyloenchondrodysplasia with immune dysregulation OMIM
ADA	NM_000022.2	100%	Severe combined immunodeficiency due to ADA deficiency OMIM Adenosine deaminase deficiency, partial OMIM
ADAR	NM_001111.4	100%	Dyschromatosis symmetrica hereditaria OMIM Aicardi-Goutieres syndrome 6 OMIM
AICDA	NM_020661.2	100%	Immunodeficiency with hyper-IgM, type 2 OMIM
AIRE	NM_000383.3	100%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia OMIM
AK2	NM_001625.3	99%	Reticular dysgenesis OMIM
AP3B1	NM_003664.4	99%	Hermansky-Pudlak syndrome 2 OMIM
ATM	NM_000051.3	100%	Ataxia-telangiectasia OMIM
B2M	NM_004048.2	100%	Immunodeficiency 43 OMIM
BCL10	NM_003921.4	100%	?Immunodeficiency 37 OMIM
BLM	NM_000057.3	100%	Bloom syndrome OMIM
BLNK	NM_013314.3	100%	Agammaglobulinemia 4 OMIM
BLOC1S6	NM_012388.2	100%	Hermansky-pudlak syndrome 9 OMIM
BRCA2	NM_000059.3	99%	Fanconi anemia, complementation group D1 OMIM

Gen	Transkript	>10x	Fenotype
BRIP1	NM_032043.2	100%	Fanconi anemia, complementation group J OMIM
BTK	NM_000061.2	100%	Agammaglobulinemia, X-linked 1 OMIM Agammaglobulinemia and isolated hormone deficiency OMIM
C15orf41	NM_001130010.2	100%	Dyserythropoietic anemia, congenital, type Ib OMIM
C1QA	NM_015991.2	100%	C1q deficiency OMIM
C1QB	NM_000491.3	100%	C1q deficiency OMIM
C1QC	NM_172369.3	100%	C1q deficiency OMIM
C1S	NM_201442.2	100%	C1s deficiency OMIM
C2	NM_000063.5	100%	C2 deficiency OMIM
C3	NM_000064.3	100%	C3 deficiency OMIM
C5	NM_001735.2	99%	C5 deficiency OMIM
C6	NM_000065.3	100%	C6 deficiency OMIM
C7	NM_000587.2	100%	C7 deficiency OMIM
C8A	NM_000562.2	100%	C8 deficiency, type I OMIM
C8B	NM_000066.3	100%	C8 deficiency, type II OMIM
C9	NM_001737.3	100%	C9 deficiency OMIM
CA2	NM_000067.2	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis OMIM
CARD11	NM_032415.5	100%	Immunodeficiency 11 OMIM
CARD9	NM_052813.4	100%	Candidiasis, familial, 2, autosomal recessive OMIM

Gen	Transkript	>10x	Fenotype
CASP10	NM_032977.3	100%	Autoimmune lymphoproliferative syndrome, type II OMIM
CASP8	NM_001228.4	100%	?Autoimmune lymphoproliferative syndrome, type IIB OMIM
CD19	NM_001770.5	100%	Immunodeficiency, common variable, 3 OMIM
CD247	NM_198053.2	100%	?Immunodeficiency 25 OMIM
CD27	NM_001242.4	100%	Lymphoproliferative syndrome 2 OMIM
CD3D	NM_000732.4	100%	Immunodeficiency 19 OMIM
CD3E	NM_000733.3	100%	Immunodeficiency 18, SCID variant OMIM Immunodeficiency 18 OMIM
CD3G	NM_000073.2	100%	Immunodeficiency 17, CD3 gamma deficient OMIM
CD40	NM_001250.5	100%	Immunodeficiency with hyper-IgM, type 3 OMIM
CD40LG	NM_000074.2	99%	Immunodeficiency, X-linked, with hyper-IgM OMIM
CD79A	NM_001783.3	100%	Agammaglobulinemia 3 OMIM
CD79B	NM_000626.2	100%	Agammaglobulinemia 6 OMIM
CD81	NM_004356.3	100%	Immunodeficiency, common variable, 6 OMIM
CD8A	NM_001768.6	100%	CD8 deficiency, familial OMIM
CDAN1	NM_138477.2	100%	Dyserythropoietic anemia, congenital, type Ia OMIM
CEBPE	NM_001805.3	100%	Specific granule deficiency OMIM
CFB	NM_001710.5	100%	?Complement factor B deficiency OMIM
CFD	NM_001928.2	98%	Complement factor D deficiency OMIM

Gen	Transkript	>10x	Fenotype
CFH	NM_000186.3	100%	Complement factor H deficiency OMIM
CFHR5	NM_030787.3	100%	Nephropathy due to CFHR5 deficiency OMIM
CFI	NM_000204.3	100%	Complement factor I deficiency OMIM
CFP	NM_002621.2	99%	PROPERDIN DEFICIENCY, X-LINKED OMIM
CHD7	NM_017780.3	100%	CHARGE syndrome OMIM
CIITA	NM_000246.3	100%	BARE LYMPHOCYTE SYNDROME, TYPE II OMIM
CLCN7	NM_001287.5	100%	Osteopetrosis, autosomal recessive 4 OMIM Osteopetrosis, autosomal dominant 2 OMIM
CLEC7A	NM_197947.2	100%	Candidiasis, familial, 4, autosomal recessive OMIM
COLEC11	NM_024027.4	100%	3MC syndrome 2 OMIM
COPA	NM_004371.3	100%	{Autoimmune interstitial lung, joint, and kidney disease} OMIM
CORO1A	NM_007074.3	92%	Immunodeficiency 8 OMIM
CR2	NM_001006658.2	100%	Immunodeficiency, common variable, 7 OMIM
CSF2RA	NM_006140.4	100%	Surfactant metabolism dysfunction, pulmonary, 4 OMIM
CSF2RB	NM_000395.2	100%	Surfactant metabolism dysfunction, pulmonary, 5 OMIM
CSF3R	NM_000760.3	99%	?Neutrophilia, hereditary OMIM
CTC1	NM_025099.5	100%	Cerebroretinal microangiopathy with calcifications and cysts OMIM
CTLA4	NM_005214.4	100%	Autoimmune lymphoproliferative syndrome, type V OMIM
CTPS1	NM_001905.3	100%	Immunodeficiency 24 OMIM

Gen	Transkript	>10x	Fenotype
CTSC	NM_001814.4	100%	Haim-Munk syndrome OMIM Periodontitis 1, juvenile OMIM
CXCR4	NM_003467.2	100%	WHIM syndrome OMIM
CYBA	NM_000101.3	98%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA OMIM
CYBB	NM_000397.3	99%	Immunodeficiency 34, mycobacteriosis, X-linked OMIM Chronic granulomatous disease, X-linked OMIM
DCLRE1C	NM_001033855.2	100%	Severe combined immunodeficiency, Athabascan type OMIM Omenn syndrome OMIM
DKC1	NM_001363.4	100%	Dyskeratosis congenita, X-linked OMIM
DNMT3B	NM_006892.3	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1 OMIM
DOCK2	NM_004946.2	100%	Immunodeficiency 40 OMIM
DOCK8	NM_203447.3	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive OMIM
DTNBP1	NM_032122.4	100%	Hermansky-Pudlak syndrome 7 OMIM
ELANE	NM_001972.2	100%	Neutropenia, cyclic OMIM Neutropenia, severe congenital 1, autosomal dominant OMIM
EPG5	NM_020964.2	99%	Vici syndrome OMIM
ERCC2	NM_000400.3	100%	Trichothiodystrophy 1, photosensitive OMIM
ERCC3	NM_000122.1	100%	Trichothiodystrophy 2, photosensitive OMIM
ERCC4	NM_005236.2	100%	Fanconi anemia, complementation group Q OMIM
ERCC6L2	NM_001010895.2	100%	Bone marrow failure syndrome 2 OMIM
F12	NM_000505.3	100%	Factor XII deficiency OMIM
FADD	NM_003824.3	100%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations OMIM
FANCA	NM_000135.2	100%	Fanconi anemia, complementation group A OMIM

Gen	Transkript	>10x	Fenotype
FANCB	NM_001018113.1	99%	Fanconi anemia, complementation group B OMIM
FANCC	NM_000136.2	100%	Fanconi anemia, complementation group C OMIM
FANCD2	NM_033084.3	100%	Fanconi anemia, complementation group D2 OMIM
FANCE	NM_021922.2	98%	Fanconi anemia, complementation group E OMIM
FANCF	NM_022725.3	100%	Fanconi anemia, complementation group F OMIM
FANCG	NM_004629.1	100%	Fanconi anemia, complementation group G OMIM
FANCI	NM_001113378.1	100%	Fanconi anemia, complementation group I OMIM
FANCL	NM_018062.3	100%	Fanconi anemia, complementation group L OMIM
FAS	NM_000043.4	100%	{Autoimmune lymphoproliferative syndrome} OMIM Autoimmune lymphoproliferative syndrome, type IA OMIM
FASLG	NM_000639.2	100%	Autoimmune lymphoproliferative syndrome, type IB OMIM
FCGR3A	NM_000569.6	100%	Immunodeficiency 20 OMIM
FCN3	NM_003665.2	100%	Immunodeficiency due to ficolin 3 deficiency OMIM
FERMT3	NM_031471.5	100%	Leukocyte adhesion deficiency, type III OMIM
FOXN1	NM_003593.2	100%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy OMIM
FOXP3	NM_014009.3	100%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked OMIM
G6PC	NM_000151.3	100%	Glycogen storage disease Ia OMIM
G6PC3	NM_138387.3	100%	Neutropenia, severe congenital 4, autosomal recessive OMIM Dursun syndrome OMIM
G6PD	NM_001042351.2	100%	Hemolytic anemia due to G6PD deficiency OMIM

Gen	Transkript	>10x	Fenotype
GATA1	NM_002049.3	100%	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities OMIM
GATA2	NM_032638.4	100%	Emberger syndrome OMIM Immunodeficiency 21 OMIM
GFI1	NM_005263.3	100%	Neutropenia, nonimmune chronic idiopathic, of adults OMIM Neutropenia, severe congenital 2, autosomal dominant OMIM
GTF2H5	NM_207118.2	100%	Trichothiodystrophy 3, photosensitive OMIM
HAX1	NM_006118.3	100%	Neutropenia, severe congenital 3, autosomal recessive OMIM
HPS1	NM_000195.3	100%	Hermansky-Pudlak syndrome 1 OMIM
HPS3	NM_032383.3	100%	Hermansky-Pudlak syndrome 3 OMIM
HPS4	NM_022081.5	100%	Hermansky-Pudlak syndrome 4 OMIM
HPS5	NM_181507.1	100%	Hermansky-Pudlak syndrome 5 OMIM
HPS6	NM_024747.5	100%	Hermansky-Pudlak syndrome 6 OMIM
ICOS	NM_012092.3	100%	Immunodeficiency, common variable, 1 OMIM
IFIH1	NM_022168.3	100%	Singleton-Merten syndrome 1 OMIM Aicardi-Goutieres syndrome 7 OMIM
IFNGR1	NM_000416.2	100%	Immunodeficiency 27A, mycobacteriosis, AR OMIM
IFNGR2	NM_005534.3	97%	Immunodeficiency 28, mycobacteriosis OMIM
IGLL1	NM_020070.3	100%	Agammaglobulinemia 2 OMIM
IKBKB	NM_001556.2	100%	Immunodeficiency 15 OMIM
IKZF1	NM_006060.5	100%	Immunodeficiency, common variable, 13 OMIM
IL10	NM_000572.2	100%	KAN GI ALVORLIG IMMUNSVIKT MED TARMAFFEKSJON

Gen	Transkript	>10x	Fenotype
IL10RA	NM_001558.3	100%	Inflammatory bowel disease 28, early onset, autosomal recessive OMIM
IL10RB	NM_000628.4	100%	INFLAMMATORY BOWEL DISEASE, EARLY-ONSET, AUTOSOMAL RECESSIVE OMIM
IL12B	NM_002187.2	100%	Immunodeficiency 29, mycobacteriosis OMIM
IL12RB1	NM_005535.2	100%	Immunodeficiency 30 OMIM
IL17F	NM_052872.3	100%	?Candidiasis, familial, 6, autosomal dominant OMIM
IL17RA	NM_014339.6	100%	?Candidiasis, familial, 5, autosomal recessive OMIM
IL17RC	NM_032732.5	100%	Candidiasis, familial, 9 OMIM
IL1RN	NM_173841.2	100%	Interleukin 1 receptor antagonist deficiency OMIM
IL21	NM_021803.3	100%	?Immunodeficiency, common variable, 11 OMIM
IL21R	NM_021798.3	100%	Immunodeficiency, primary, autosomal recessive, IL21R-related OMIM
IL2RA	NM_000417.2	100%	Immunodeficiency 41 with lymphoproliferation and autoimmunity OMIM
IL2RG	NM_000206.2	100%	Severe combined immunodeficiency, X-linked OMIM Combined immunodeficiency, X-linked, moderate OMIM
IL36RN	NM_012275.2	100%	Psoriasis 14, pustular OMIM
IL7R	NM_002185.3	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type OMIM
IRAK4	NM_016123.3	100%	Invasive pneumococcal disease, recurrent isolated, 1 OMIM IRAK4 deficiency OMIM
IRF7	NM_004031.2	100%	?Immunodeficiency 39 OMIM
IRF8	NM_002163.2	100%	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive OMIM Immunodeficiency 32A, mycobacteriosis, autosomal dominant OMIM
ISG15	NM_005101.3	100%	Immunodeficiency 38 OMIM

Gen	Transkript	>10x	Fenotype
ITCH	NM_031483.5	100%	Autoimmune disease, multisystem, with facial dysmorphism OMIM
ITGB2	NM_000211.4	100%	Leukocyte adhesion deficiency OMIM
ITK	NM_005546.3	100%	Lymphoproliferative syndrome 1 OMIM
JAGN1	NM_032492.3	100%	Neutropenia, severe congenital, 6, autosomal recessive OMIM
JAK3	NM_000215.3	99%	SCID, autosomal recessive, T-negative/B-positive type OMIM
KMT2D	NM_003482.3	100%	Kabuki syndrome 1 OMIM
LAMTOR2	NM_014017.3	100%	Immunodeficiency due to defect in MAPBP-interacting protein OMIM
LCK	NM_001042771.2	100%	?Immunodeficiency 22 OMIM
LIG4	NM_002312.3	100%	LIG4 syndrome OMIM
LPIN2	NM_014646.2	100%	Majeed syndrome OMIM
LRBA	NM_006726.4	100%	Immunodeficiency, common variable, 8, with autoimmunity OMIM
LRRC8A	NM_019594.3	100%	?Agammaglobulinemia 5 OMIM
LYST	NM_000081.3	99%	Chediak-Higashi syndrome OMIM
MAGT1	NM_032121.5	100%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia OMIM
MALT1	NM_006785.3	92%	Immunodeficiency 12 OMIM
MASP2	NM_006610.3	100%	MASP2 deficiency OMIM
MBL2	NM_000242.2	100%	{Chronic infections, due to MBL deficiency} OMIM
MBTPS2	NM_015884.3	100%	?Olmsted syndrome, X-linked OMIM

Gen	Transkript	>10x	Fenotype
MCM4	NM_005914.3	100%	Natural killer cell and glucocorticoid deficiency with DNA repair defect OMIM
MEFV	NM_000243.2	100%	Familial Mediterranean fever, AR OMIM Familial Mediterranean fever, AD OMIM
MLPH	NM_024101.6	100%	Griscelli syndrome, type 3 OMIM
MPO	NM_000250.1	100%	Myeloperoxidase deficiency OMIM
MRE11A	NM_005591.3	100%	Ataxia-telangiectasia-like disorder OMIM
MS4A1	NM_021950.3	100%	Immunodeficiency, common variable, 5 OMIM
MTHFD1	NM_005956.3	100%	SCID (+megaloblastic anemi, HUS og div andre, kan behandles med FOLAT) PubMed SCID (+megaloblastic anemi, HUS og div andre, kan behandles med FOLAT) PubMed
MVK	NM_000431.3	100%	Mevalonic aciduria OMIM Hyper-IgD syndrome OMIM
MYD88	NM_002468.4	100%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency OMIM
MYH9	NM_002473.5	100%	Fechtner syndrome OMIM Epstein syndrome OMIM Sebastian syndrome OMIM May-Hegglin anomaly OMIM Macrothrombocytopenia and progressive sensorineural deafness OMIM
MYO5A	NM_000259.3	100%	Griscelli syndrome, type 1 OMIM
NBN	NM_002485.4	100%	Nijmegen breakage syndrome OMIM
NCF2	NM_000433.3	100%	Chronic granulomatous disease due to deficiency of NCF-2 OMIM
NCF4	NM_013416.3	100%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III OMIM
NFKB1	NM_003998.3	100%	Immunodeficiency, common variable, 12 OMIM
NFKB2	NM_001077494.3	100%	Immunodeficiency, common variable, 10 OMIM
NFKBIA	NM_020529.2	99%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency OMIM

Gen	Transkript	>10x	Fenotype
NHEJ1	NM_024782.2	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation OMIM
NHP2	NM_017838.3	100%	Dyskeratosis congenita, autosomal recessive 2 OMIM
NLRC4	NM_001199139.1	100%	Autoinflammation with infantile enterocolitis OMIM ?Familial cold autoinflammatory syndrome 4 OMIM
NLRP1	NM_033004.3	100%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia OMIM
NLRP12	NM_144687.3	100%	Familial cold autoinflammatory syndrome 2 OMIM
NLRP3	NM_004895.4	100%	Muckle-Wells syndrome OMIM Familial cold-induced inflammatory syndrome 1 OMIM CINCA syndrome OMIM
NOD2	NM_022162.2	100%	Blau syndrome OMIM
NOP10	NM_018648.3	100%	Dyskeratosis congenita, autosomal recessive 1 OMIM
NRAS	NM_002524.4	100%	Noonan syndrome 6 OMIM
ORAI1	NM_032790.3	99%	Immunodeficiency 9 OMIM
OSTM1	NM_014028.3	98%	Osteopetrosis, autosomal recessive 5 OMIM
PALB2	NM_024675.3	100%	Fanconi anemia, complementation group N OMIM
PARN	NM_002582.3	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4 OMIM
PGM3	NM_001199917.1	100%	Immunodeficiency 23 OMIM
PIEZO1	NM_001142864.2	100%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema OMIM
PIK3CD	NM_005026.3	99%	Immunodeficiency 14 OMIM
PIK3R1	NM_181523.2	100%	Immunodeficiency 36 OMIM ?Agammaglobulinemia 7, autosomal recessive OMIM
PLCG2	NM_002661.4	100%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome OMIM

Gen	Transkript	>10x	Fenotype
PLEKHM1	NM_014798.2	99%	Osteopetrosis, autosomal recessive 6 OMIM
PNP	NM_000270.3	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency OMIM
POLE	NM_006231.3	100%	FILS syndrome OMIM
PRF1	NM_001083116.1	100%	Aplastic anemia OMIM
PRKCD	NM_006254.3	100%	Autoimmune lymphoproliferative syndrome, type III OMIM
PRKDC	NM_006904.6	99%	Immunodeficiency 26, with or without neurologic abnormalities OMIM
PSMB8	NM_148919.3	100%	Autoinflammation, lipodystrophy, and dermatosis syndrome OMIM
PSTPIP1	NM_003978.3	100%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne OMIM
PTPRC	NM_002838.4	99%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive OMIM
RAB27A	NM_004580.4	100%	Griscelli syndrome, type 2 OMIM
RAC2	NM_002872.4	100%	Neutrophil immunodeficiency syndrome OMIM
RAD50	NM_005732.3	99%	Nijmegen breakage syndrome-like disorder OMIM
RAD51C	NM_058216.2	100%	Fanconi anemia, complementation group O OMIM
RAG1	NM_000448.2	100%	Severe combined immunodeficiency, B cell-negative OMIM Omenn syndrome OMIM Combined cellular and humoral immune defects with granulomas OMIM Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity OMIM
RAG2	NM_000536.3	100%	Omenn syndrome OMIM Combined cellular and humoral immune defects with granulomas OMIM Severe combined immunodeficiency, B cell-negative OMIM
RASGRP2	NM_153819.1	100%	?Bleeding disorder, platelet-type, 18 OMIM
RBCK1	NM_031229.2	99%	Polyglucosan body myopathy 1 with or without immunodeficiency OMIM

Gen	Transkript	>10x	Fenotype
RBM8A	NM_005105.4	100%	Thrombocytopenia-absent radius syndrome OMIM
RECQL4	NM_004260.3	99%	Rothmund-Thomson syndrome OMIM RAPADILINO syndrome OMIM Baller-Gerold syndrome OMIM PubMed
RFX5	NM_000449.3	100%	Bare lymphocyte syndrome, type II, complementation group E OMIM Bare lymphocyte syndrome, type II, complementation group C OMIM
RFXANK	NM_003721.3	100%	MHC class II deficiency, complementation group B OMIM
RFXAP	NM_000538.3	100%	Bare lymphocyte syndrome, type II, complementation group D OMIM
RHAG	NM_000324.2	100%	Overhydrated hereditary stomatocytosis OMIM Anemia, hemolytic, Rh-null, regulator type OMIM
RHOH	NM_004310.4	100%	Kronisk epidermodysplasia verruciformis/ øket disposisjon for HPV PubMed
RLTPR	NM_001013838.1	99%	IN PRESS ASP: 4 pasienter i 3 norske fam. med mutasjon i genet
RNASEH2A	NM_006397.2	100%	Aicardi-Goutieres syndrome 4 OMIM
RNASEH2B	NM_024570.3	100%	Aicardi-Goutieres syndrome 2 OMIM
RNASEH2C	NM_032193.3	100%	Aicardi-Goutieres syndrome 3 OMIM
RNF168	NM_152617.3	100%	RIDDLE syndrome OMIM
RNF31	NM_017999.4	100%	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia PubMed
RORC	NM_005060.3	100%	Immunodeficiency 42 OMIM
RPL11	NM_000975.3	100%	Diamond-Blackfan anemia 7 OMIM
RPL15	NM_001253384.1	81%	?Diamond-Blackfan anemia 12 OMIM
RPL26	NM_000987.3	100%	?Diamond-Blackfan anemia 11 OMIM
RPL35A	NM_000996.2	99%	Diamond-Blackfan anemia 5 OMIM

Gen	Transkript	>10x	Fenotype
RPL5	NM_000969.3	96%	Diamond-Blackfan anemia 6 OMIM
RPS10	NM_001014.4	100%	Diamond-Blackfan anemia 9 OMIM
RPS19	NM_001022.3	100%	Diamond-Blackfan anemia 1 OMIM
RPS24	NM_033022.3	99%	Diamond-blackfan anemia 3 OMIM
RPS26	NM_001029.3	99%	Diamond-Blackfan anemia 10 OMIM
RPS29	NM_001032.4	100%	Diamond-Blackfan anemia 13 OMIM
RPS7	NM_001011.3	98%	Diamond-Blackfan anemia 8 OMIM
RPSA	NM_002295.5	100%	Asplenia, isolated congenital OMIM
RTEL1	NM_032957.4	99%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3 OMIM
RUNX1	NM_001754.4	100%	Platelet disorder, familial, with associated myeloid malignancy OMIM
SAMHD1	NM_015474.3	100%	Aicardi-Goutieres syndrome 5 OMIM
SBDS	NM_016038.2	100%	Shwachman-Diamond syndrome OMIM
SERPING1	NM_000062.2	100%	Complement component 4, partial deficiency of OMIM
SH2D1A	NM_002351.4	100%	Lymphoproliferative syndrome, X-linked, 1 OMIM
SKIV2L	NM_006929.4	100%	Trichohepatoenteric syndrome 2 OMIM
SLC29A3	NM_018344.5	100%	Histiocytosis-lymphadenopathy plus syndrome OMIM
SLC35C1	NM_018389.4	100%	Congenital disorder of glycosylation, type IIc OMIM
SLC37A4	NM_001164277.1	99%	Glycogen storage disease Ib OMIM Glycogen storage disease Ic OMIM

Gen	Transkript	>10x	Fenotype
SLC46A1	NM_080669.5	99%	Folate malabsorption, hereditary OMIM
SLX4	NM_032444.2	100%	Fanconi anemia, complementation group P OMIM
SMARCAL1	NM_014140.3	100%	Schimke immunoosseous dysplasia OMIM
SNX10	NM_001199835.1	100%	Osteopetrosis, autosomal recessive 8 OMIM
SP110	NM_004509.3	100%	Hepatic venoocclusive disease with immunodeficiency OMIM
SPINK5	NM_006846.3	100%	Netherton syndrome OMIM Atopy OMIM
SRP72	NM_006947.3	99%	Bone marrow failure syndrome 1 OMIM
STAT1	NM_007315.3	99%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant OMIM Immunodeficiency 31C, autosomal dominant OMIM Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive OMIM
STAT2	NM_005419.3	100%	Immunodeficiency 44 OMIM
STAT3	NM_139276.2	100%	Hyper-IgE recurrent infection syndrome OMIM Autoimmune disease, multisystem, infantile-onset OMIM
STAT5B	NM_012448.3	96%	Growth hormone insensitivity with immunodeficiency OMIM
STIM1	NM_003156.3	99%	Immunodeficiency 10 OMIM
STK4	NM_006282.2	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations OMIM
STX11	NM_003764.3	100%	Hemophagocytic lymphohistiocytosis, familial, 4 OMIM
STXBP2	NM_006949.3	100%	Hemophagocytic lymphohistiocytosis, familial, 5 OMIM
TAP1	NM_000593.5	100%	Bare lymphocyte syndrome, type I OMIM
TAP2	NM_000544.3	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency OMIM
TAPBP	NM_003190.4	100%	Bare lymphocyte syndrome, type I OMIM

Gen	Transkript	>10x	Fenotype
TAZ	NM_000116.4	100%	Barth syndrome OMIM
TBX1	NM_080647.1	86%	DiGeorge syndrome OMIM Velocardiofacial syndrome OMIM
TCF3	NM_003200.3	99%	Agammaglobulinemia 8, autosomal dominant OMIM
TCIRG1	NM_006019.3	99%	Osteopetrosis, autosomal recessive 1 OMIM
TCN2	NM_000355.3	100%	Transcobalamin II deficiency OMIM
TERT	NM_198253.2	99%	{Dyskeratosis congenita, autosomal recessive 4} OMIM {Dyskeratosis congenita, autosomal dominant 2} OMIM {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1} OMIM
TFRC	NM_003234.2	99%	Immunodeficiency 46 OMIM
TGFB1	NM_000660.5	100%	Camurati-Engelmann disease OMIM
THBD	NM_000361.2	100%	{Hemolytic uremic syndrome, atypical, susceptibility to, 6} OMIM
TICAM1	NM_182919.3	100%	{Herpes simplex encephalitic, susceptibility to, 6} OMIM
TINF2	NM_001099274.1	100%	Dyskeratosis congenita, autosomal dominant 3 OMIM Revesz syndrome OMIM
TLR3	NM_003265.2	100%	{Herpes simplex encephalitis, susceptibility to, 2} OMIM
TMC6	NM_007267.6	99%	Epidermodysplasia verruciformis OMIM
TMC8	NM_152468.4	99%	Epidermodysplasia verruciformis OMIM
TMEM173	NM_198282.3	100%	STING-associated vasculopathy, infantile-onset OMIM
TNFRSF11A	NM_003839.3	97%	Osteopetrosis, autosomal recessive 7 OMIM
TNFRSF13B	NM_012452.2	100%	Immunodeficiency, common variable, 2 OMIM Immunoglobulin A deficiency 2 OMIM
TNFRSF13C	NM_052945.3	100%	Immunodeficiency, common variable, 4 OMIM

Gen	Transkript	>10x	Fenotype
TNFRSF1A	NM_001065.3	100%	Periodic fever, familial OMIM
TNFRSF4	NM_003327.3	99%	?Immunodeficiency 16 OMIM
TNFSF11	NM_003701.3	100%	Osteopetrosis, autosomal recessive 2 OMIM
TNFSF12	NM_003809.2	99%	B celle svikt/ immunglobulinmangel/ nedsatt humoralt immunrespons PubMed
TPP2	NM_003291.2	100%	Early-onset Evans syndrome, immunodeficiency, and premature immunosenescence associated with tripeptidyl-peptidase II deficiency PubMed
TRAF3	NM_003300.3	100%	{?Herpes simplex encephalitis, susceptibility to, 3} OMIM
TRAF3IP2	NM_147686.3	100%	?Candidiasis, familial, 8 OMIM
TREX1	NM_033629.4	100%	Aicardi-Goutieres syndrome 1, dominant and recessive OMIM
TRNT1	NM_182916.2	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay OMIM
TTC37	NM_014639.3	100%	Trichohepatoenteric syndrome 1 OMIM
TTC7A	NM_020458.3	100%	Gastrointestinal defects and immunodeficiency syndrome OMIM
TYK2	NM_003331.4	100%	Immunodeficiency 35 OMIM
UNC119	NM_005148.3	100%	?Immunodeficiency 13 OMIM
UNC13D	NM_199242.2	99%	Hemophagocytic lymphohistiocytosis, familial, 3 OMIM
UNG	NM_080911.2	100%	Immunodeficiency with hyper IgM, type 5 OMIM
USB1	NM_024598.3	100%	Poikiloderma med neutropeni OMIM
VPS13B	NM_017890.4	100%	Cohen syndrome OMIM
VPS45	NM_007259.4	100%	Neutropenia, severe congenital, 5, autosomal recessive OMIM

Gen	Transkript	>10x	Fenotype
WAS	NM_000377.2	91%	Neutropenia, severe congenital, X-linked OMIM Wiskott-Aldrich syndrome OMIM Thrombocytopenia, X-linked, intermittent OMIM Thrombocytopenia, X-linked OMIM
WIPF1	NM_001077269.1	100%	?Wiskott-Aldrich syndrome 2 OMIM
WRAP53	NM_018081.2	100%	Dyskeratosis congenita, autosomal recessive 3 OMIM
XIAP	NM_001167.3	94%	Lymphoproliferative syndrome, X-linked, 2 OMIM
ZAP70	NM_001079.3	100%	Selective T-cell defect OMIM
ZBTB24	NM_014797.2	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2 OMIM