Oslo universitetssykehus
Avdeling for medisinsk genetikk

Immunsviktsykdommer

Genpanel, versjon v03

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet , gjenkjennes (https://genome.ucsc.edu).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner, så disse vil ikke bli analysert: **CSF2RA, FCGR3A, PLEKHM1, RBM8A, RPL15, SBDS**

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ACD	25070	NM_001082487.1		1-12	?Dyskeratosis congenita, autosomal recessive 7 OMIM ?Dyskeratosis congenita, autosomal dominant 6 OMIM
ACP5	124	NM_001111035.1		4-7	Spondyloenchondrodysplasia with immune dysregulation OMIM
ADA	186	NM_000022.2		1-12	Severe combined immunodeficiency due to ADA deficiency OMIM Adenosine deaminase deficiency, partial OMIM
ADAR	225	NM_001111.4		1-15	Dyschromatosis symmetrica hereditaria OMIM Aicardi-Goutieres syndrome 6 OMIM
AICDA	13203	NM_020661.2		1-5	Immunodeficiency with hyper-IgM, type 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
AIRE	360	NM_000383.3		1-14	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia OMIM
AK2	362	NM_001625.3		1-6	Reticular dysgenesis OMIM
AP3B1	566	NM_003664.4		1-27	Hermansky-Pudlak syndrome 2 OMIM
ATM	795	NM_000051.3		2-63	Ataxia-telangiectasia OMIM
B2M	914	NM_004048.2		1-3	Immunodeficiency 43 OMIM
BCL10	989	NM_003921.4		1-3	?Immunodeficiency 37 OMIM
BLM	1058	NM_000057.3		2-22	Bloom syndrome OMIM
BLNK	14211	NM_013314.3		1-17	Agammaglobulinemia 4 OMIM
BLOC1S6	8549	NM_012388.2		1-5	Hermansky-pudlak syndrome 9 OMIM
BRCA2	1101	NM_000059.3		2-27	Fanconi anemia, complementation group D1 OMIM
BRIP1	20473	NM_032043.2		2-20	Fanconi anemia, complementation group J OMIM
BTK	1133	NM_000061.2		2-19	Agammaglobulinemia, X-linked 1 OMIM Agammaglobulinemia and isolated hormone deficiency OMIM
C15orf41	26929	NM_001130010.2		1-11	Dyserythropoietic anemia, congenital, type Ib OMIM
C1QA	1241	NM_015991.2		2-3	C1q deficiency OMIM
C1QB	1242	NM_000491.3		2-3	C1q deficiency OMIM

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C1QC	1245	NM_172369.3		2-3	C1q deficiency OMIM
C1S	1247	NM_201442.2		2-12	C1s deficiency OMIM
C2	1248	NM_000063.5		1-18	C2 deficiency OMIM
C3	1318	NM_000064.3		1-41	C3 deficiency OMIM
C5	1331	NM_001735.2		1-41	C5 deficiency OMIM
C6	1339	NM_000065.3		2-18	C6 deficiency OMIM
C7	1346	NM_000587.2		1-18	C7 deficiency OMIM
C8A	1352	NM_000562.2		1-11	C8 deficiency, type I OMIM
C8B	1353	NM_000066.3		1-12	C8 deficiency, type II OMIM
C9	1358	NM_001737.3		1-11	C9 deficiency OMIM
CA2	1373	NM_000067.2		1-7	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis OMIM
CARD11	16393	NM_032415.5		2-25	Immunodeficiency 11 OMIM
CARD9	16391	NM_052813.4		2-13	Candidiasis, familial, 2, autosomal recessive OMIM
CASP10	1500	NM_032977.3		2-10	Autoimmune lymphoproliferative syndrome, type II OMIM
CASP8	1509	NM_001228.4		3-10	?Autoimmune lymphoproliferative syndrome, type IIB OMIM
CD19	1633	NM_001770.5		1-14	Immunodeficiency, common variable, 3 OMIM

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CD247	1677	NM_198053.2		1-8	?Immunodeficiency 25 OMIM
CD27	11922	NM_001242.4		1-6	Lymphoproliferative syndrome 2 OMIM
CD3D	1673	NM_000732.4		1-5	Immunodeficiency 19 OMIM
CD3E	1674	NM_000733.3		2-9	Immunodeficiency 18, SCID variant OMIM Immunodeficiency 18 OMIM
CD3G	1675	NM_000073.2		1-6	Immunodeficiency 17, CD3 gamma deficient OMIM
CD40	11919	NM_001250.5		1-9	Immunodeficiency with hyper-IgM, type 3 OMIM
CD40LG	11935	NM_000074.2		1-5	Immunodeficiency, X-linked, with hyper-IgM OMIM
CD79A	1698	NM_001783.3		1-5	Agammaglobulinemia 3 OMIM
CD79B	1699	NM_000626.2		1-6	Agammaglobulinemia 6 OMIM
CD81	1701	NM_004356.3		1-8	Immunodeficiency, common variable, 6 OMIM
CD8A	1706	NM_001768.6		1-6	CD8 deficiency, familial OMIM
CDAN1	1713	NM_138477.2		1-28	Dyserythropoietic anemia, congenital, type Ia OMIM
CEBPE	1836	NM_001805.3		1-2	Specific granule deficiency OMIM
CFB	1037	NM_001710.5		1-18	?Complement factor B deficiency OMIM
CFD	2771	NM_001928.2		1-5	Complement factor D deficiency OMIM
CFH	4883	NM_000186.3	8-10, 20-22	1-22	Complement factor H deficiency OMIM

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CFHR5	24668	NM_030787.3		1-10	Nephropathy due to CFHR5 deficiency OMIM
CFI	5394	NM_000204.3		1-13	Complement factor I deficiency OMIM
CFP	8864	NM_002621.2		2-10	PROPERDIN DEFICIENCY, X-LINKED OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM
CIITA	7067	NM_000246.3		1-19	BARE LYMPHOCYTE SYNDROME, TYPE II OMIM
CLCN7	2025	NM_001287.5		1-25	Osteopetrosis, autosomal recessive 4 OMIM Osteopetrosis, autosomal dominant 2 OMIM
CLEC7A	14558	NM_197947.2		1-6	Candidiasis, familial, 4, autosomal recessive OMIM
COLEC11	17213	NM_024027.4		2-7	3MC syndrome 2 OMIM
COPA	2230	NM_004371.3		1-33	{Autoimmune interstitial lung, joint, and kidney disease} OMIM
CORO1A	2252	NM_007074.3	10-11	2-11	Immunodeficiency 8 OMIM
CR2	2336	NM_001006658.2		1-19	Immunodeficiency, common variable, 7 OMIM
CSF2RA	2435	NM_006140.4	3-13	3-13	Surfactant metabolism dysfunction, pulmonary, 4 OMIM
CSF2RB	2436	NM_000395.2		2-14	Surfactant metabolism dysfunction, pulmonary, 5 OMIM
CSF3R	2439	NM_000760.3		3-17	?Neutrophilia, hereditary OMIM
CTC1	26169	NM_025099.5		1-23	Cerebroretinal microangiopathy with calcifications and cysts OMIM
CTLA4	2505	NM_005214.4		1-4	Autoimmune lymphoproliferative syndrome, type V OMIM

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CTPS1	2519	NM_001905.3		2-18	Immunodeficiency 24 OMIM
CTSC	2528	NM_001814.4		1-7	Haim-Munk syndrome OMIM Periodontitis 1, juvenile OMIM
CXCR4	2561	NM_003467.2		1-2	WHIM syndrome OMIM
CYBA	2577	NM_000101.3		1-6	Chronic granulomatous disease, autosomal, due to deficiency of CYBA OMIM
CYBB	2578	NM_000397.3		1-13	Immunodeficiency 34, mycobacteriosis, X-linked OMIM Chronic granulomatous disease, X-linked OMIM
DCLRE1C	17642	NM_001033855.2	4-9	1-14	Severe combined immunodeficiency, Athabascan type OMIM Omenn syndrome OMIM
DKC1	2890	NM_001363.4		1-15	Dyskeratosis congenita, X-linked OMIM
DNMT3B	2979	NM_006892.3		2-23	Immunodeficiency-centromeric instability-facial anomalies syndrome 1 OMIM
DOCK2	2988	NM_004946.2		1-52	Immunodeficiency 40 OMIM
DOCK8	19191	NM_203447.3		1-48	Hyper-IgE recurrent infection syndrome, autosomal recessive OMIM
DTNBP1	17328	NM_032122.4		1-10	Hermansky-Pudlak syndrome 7 OMIM
ELANE	3309	NM_001972.2		1-5	Neutropenia, cyclic OMIM Neutropenia, severe congenital 1, autosomal dominant OMIM
EPG5	29331	NM_020964.2		1-44	Vici syndrome OMIM
ERCC2	3434	NM_000400.3		1-23	Trichothiodystrophy 1, photosensitive OMIM

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ERCC3	3435	NM_000122.1		1-15	Trichothiodystrophy 2, photosensitive OMIM
ERCC4	3436	NM_005236.2		1-11	Fanconi anemia, complementation group Q OMIM
ERCC6L2	26922	NM_001010895.2		1-14	Bone marrow failure syndrome 2 OMIM
F12	3530	NM_000505.3		1-14	Factor XII deficiency OMIM
FADD	3573	NM_003824.3		1-2	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations OMIM
FANCA	3582	NM_000135.2		1-43	Fanconi anemia, complementation group A OMIM
FANCB	3583	NM_001018113.1		3-10	Fanconi anemia, complementation group B OMIM
FANCC	3584	NM_000136.2		2-15	Fanconi anemia, complementation group C OMIM
FANCD2	3585	NM_033084.3	12-17, 19-28	2-43	Fanconi anemia, complementation group D2 OMIM
FANCE	3586	NM_021922.2		1-10	Fanconi anemia, complementation group E OMIM
FANCF	3587	NM_022725.3		1	Fanconi anemia, complementation group F OMIM
FANCG	3588	NM_004629.1		1-14	Fanconi anemia, complementation group G OMIM
FANCI	25568	NM_001113378.1		2-38	Fanconi anemia, complementation group I OMIM
FANCL	20748	NM_018062.3		1-14	Fanconi anemia, complementation group L OMIM
FAS	11920	NM_000043.4		1-9	{Autoimmune lymphoproliferative syndrome} OMIM Autoimmune lymphoproliferative syndrome, type IA OMIM

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FASLG	11936	NM_000639.2		1-4	Autoimmune lymphoproliferative syndrome, type IB OMIM
FCGR3A	3619	NM_000569.6	1-5	1-5	Immunodeficiency 20 OMIM
FCN3	3625	NM_003665.2		1-8	Immunodeficiency due to ficolin 3 deficiency OMIM
FERMT3	23151	NM_031471.5		2-15	Leukocyte adhesion deficiency, type III OMIM
FOXN1	12765	NM_003593.2		1-8	T-cell immunodeficiency, congenital alopecia, and nail dystrophy OMIM
FOXP3	6106	NM_014009.3		2-12	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked OMIM
G6PC	4056	NM_000151.3		1-5	Glycogen storage disease Ia OMIM
G6PC3	24861	NM_138387.3		1-6	Neutropenia, severe congenital 4, autosomal recessive OMIM Dursun syndrome OMIM
G6PD	4057	NM_001042351.2		2-13	Hemolytic anemia due to G6PD deficiency OMIM
GATA1	4170	NM_002049.3		2-6	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities OMIM
GATA2	4171	NM_032638.4		2-6	Emberger syndrome OMIM Immunodeficiency 21 OMIM
GFI1	4237	NM_005263.3		2-7	Neutropenia, nonimmune chronic idiopathic, of adults OMIM Neutropenia, severe congenital 2, autosomal dominant OMIM
GTF2H5	21157	NM_207118.2		2-3	Trichothiodystrophy 3, photosensitive OMIM
HAX1	16915	NM_006118.3		1-7	Neutropenia, severe congenital 3, autosomal recessive OMIM
HPS1	5163	NM_000195.3	4-6	3-20	Hermansky-Pudlak syndrome 1 OMIM

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HPS3	15597	NM_032383.3		1-17	Hermansky-Pudlak syndrome 3 OMIM
HPS4	15844	NM_022081.5		2-14	Hermansky-Pudlak syndrome 4 OMIM
HPS5	17022	NM_181507.1		2-23	Hermansky-Pudlak syndrome 5 OMIM
HPS6	18817	NM_024747.5		1	Hermansky-Pudlak syndrome 6 OMIM
ICOS	5351	NM_012092.3		1-5	Immunodeficiency, common variable, 1 OMIM
IFIH1	18873	NM_022168.3		1-16	Singleton-Merten syndrome 1 OMIM Aicardi-Goutieres syndrome 7 OMIM
IFNGR1	5439	NM_000416.2		1-7	Immunodeficiency 27A, mycobacteriosis, AR OMIM
IFNGR2	5440	NM_005534.3		1-7	Immunodeficiency 28, mycobacteriosis OMIM
IGLL1	5870	NM_020070.3	2-3	1-3	Agammaglobulinemia 2 OMIM
IKBKB	5960	NM_001556.2		2-22	Immunodeficiency 15 OMIM
IKZF1	13176	NM_006060.5		2-8	Immunodeficiency, common variable, 13 OMIM
IL10	5962	NM_000572.2		1-5	KAN GI ALVORLIG IMMUNSVIKT MED TARMAFFEKSJON
IL10RA	5964	NM_001558.3		1-7	Inflammatory bowel disease 28, early onset, autosomal recessive OMIM
IL10RB	5965	NM_000628.4		1-7	INFLAMMATORY BOWEL DISEASE, EARLY-ONSET, AUTOSOMAL RECESSIVE OMIM
IL12B	5970	NM_002187.2		2-7	Immunodeficiency 29, mycobacteriosis OMIM
IL12RB1	5971	NM_005535.2		1-17	Immunodeficiency 30 OMIM

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IL17F	16404	NM_052872.3		1-3	?Candidiasis, familial, 6, autosomal dominant OMIM
IL17RA	5985	NM_014339.6		1-13	?Candidiasis, familial, 5, autosomal recessive OMIM
IL17RC	18358	NM_032732.5		1-18	Candidiasis, familial, 9 OMIM
IL1RN	6000	NM_173841.2		1-6	Interleukin 1 receptor antagonist deficiency OMIM
IL21	6005	NM_021803.3		1-5	?Immunodeficiency, common variable, 11 OMIM
IL21R	6006	NM_021798.3		2-9	Immunodeficiency, primary, autosomal recessive, IL21R-related OMIM
IL2RA	6008	NM_000417.2		1-8	Immunodeficiency 41 with lymphoproliferation and autoimmunity OMIM
IL2RG	6010	NM_000206.2		1-8	Severe combined immunodeficiency, X-linked OMIM Combined immunodeficiency, X-linked, moderate OMIM
IL36RN	15561	NM_012275.2		2-5	Psoriasis 14, pustular OMIM
IL7R	6024	NM_002185.3		1-8	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type OMIM
IRAK4	17967	NM_016123.3		2-12	Invasive pneumococcal disease, recurrent isolated, 1 OMIM IRAK4 deficiency OMIM
IRF7	6122	NM_004031.2		1-9	?Immunodeficiency 39 OMIM
IRF8	5358	NM_002163.2		2-9	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive OMIM Immunodeficiency 32A, mycobacteriosis, autosomal dominant OMIM

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ISG15	4053	NM_005101.3		1-2	Immunodeficiency 38 OMIM
ITCH	13890	NM_031483.5		3-25	Autoimmune disease, multisystem, with facial dysmorphism OMIM
ITGB2	6155	NM_000211.4		2-16	Leukocyte adhesion deficiency OMIM
ITK	6171	NM_005546.3		1-17	Lymphoproliferative syndrome 1 OMIM
JAGN1	26926	NM_032492.3		1-2	Neutropenia, severe congenital, 6, autosomal recessive OMIM
JAK3	6193	NM_000215.3		2-24	SCID, autosomal recessive, T-negative/B-positive type OMIM
KMT2D	7133	NM_003482.3		1-54	Kabuki syndrome 1 OMIM
LAMTOR2	29796	NM_014017.3		1-4	Immunodeficiency due to defect in MAPBP-interacting protein OMIM
LCK	6524	NM_001042771.2		2-13	?Immunodeficiency 22 OMIM
LIG4	6601	NM_002312.3		2	LIG4 syndrome OMIM
LPIN2	14450	NM_014646.2		2-20	Majeed syndrome OMIM
LRBA	1742	NM_006726.4		2-58	Immunodeficiency, common variable, 8, with autoimmunity OMIM
LRRC8A	19027	NM_019594.3		3-4	?Agammaglobulinemia 5 OMIM
LYST	1968	NM_000081.3		3-53	Chediak-Higashi syndrome OMIM
MAGT1	28880	NM_032121.5		1-10	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia OMIM
MALT1	6819	NM_006785.3		1-17	Immunodeficiency 12 OMIM

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MASP2	6902	NM_006610.3		1-11	MASP2 deficiency OMIM
MBL2	6922	NM_000242.2		1-4	{Chronic infections, due to MBL deficiency} OMIM
MBTPS2	15455	NM_015884.3		1-11	?Olmsted syndrome, X-linked OMIM
MCM4	6947	NM_005914.3		1-16	Natural killer cell and glucocorticoid deficiency with DNA repair defect OMIM
MEFV	6998	NM_000243.2		1-10	Familial Mediterranean fever, AR OMIM Familial Mediterranean fever, AD OMIM
MLPH	29643	NM_024101.6		2-16	Griscelli syndrome, type 3 OMIM
MPO	7218	NM_000250.1		1-12	Myeloperoxidase deficiency OMIM
MRE11A	7230	NM_005591.3		2-20	Ataxia-telangiectasia-like disorder OMIM
MS4A1	7315	NM_021950.3		2-7	Immunodeficiency, common variable, 5 OMIM
MTHFD1	7432	NM_005956.3		1-27	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	7530	NM_000431.3		2-11	Mevalonic aciduria OMIM Hyper-IgD syndrome OMIM
MYD88	7562	NM_002468.4		1-5	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency OMIM

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MYH9	7579	NM_002473.5		2-41	Fechtner syndrome OMIM Epstein syndrome OMIM Sebastian syndrome OMIM May-Hegglin anomaly OMIM Macrothrombocytopenia and progressive sensorineural deafness OMIM
MYO5A	7602	NM_000259.3		1-41	Griscelli syndrome, type 1 OMIM
NBN	7652	NM_002485.4		1-16	Nijmegen breakage syndrome OMIM
NCF2	7661	NM_000433.3		1-15	Chronic granulomatous disease due to deficiency of NCF-2 OMIM
NCF4	7662	NM_013416.3		1-8	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III OMIM
NFKB1	7794	NM_003998.3		2-24	Immunodeficiency, common variable, 12 OMIM
NFKB2	7795	NM_001077494.3		2-23	Immunodeficiency, common variable, 10 OMIM
NFKBIA	7797	NM_020529.2		1-6	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency OMIM
NHEJ1	25737	NM_024782.2		2-8	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation OMIM
NHP2	14377	NM_017838.3		1-4	Dyskeratosis congenita, autosomal recessive 2 OMIM
NLRC4	16412	NM_001199139.1		2-9	Autoinflammation with infantile enterocolitis OMIM ?Familial cold autoinflammatory syndrome 4 OMIM
NLRP1	14374	NM_033004.3		1-17	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia OMIM
NLRP12	22938	NM_144687.3		1-10	Familial cold autoinflammatory syndrome 2 OMIM

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NLRP3	16400	NM_004895.4		1-9	Muckle-Wells syndrome OMIM Familial cold-induced inflammatory syndrome 1 OMIM CINCA syndrome OMIM
NOD2	5331	NM_022162.2		1-12	Blau syndrome OMIM
NOP10	14378	NM_018648.3		1-2	Dyskeratosis congenita, autosomal recessive 1 OMIM
NRAS	7989	NM_002524.4		2-5	Noonan syndrome 6 OMIM
ORAI1	25896	NM_032790.3		1-2	Immunodeficiency 9 OMIM
OSTM1	21652	NM_014028.3		1-6	Osteopetrosis, autosomal recessive 5 OMIM
PALB2	26144	NM_024675.3		1-13	Fanconi anemia, complementation group N OMIM
PARN	8609	NM_002582.3	24	1-24	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4 OMIM
PGM3	8907	NM_001199917.1		2-14	Immunodeficiency 23 OMIM
PIEZO1	28993	NM_001142864.2		1-51	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema OMIM
PIK3CD	8977	NM_005026.3	24	3-24	Immunodeficiency 14 OMIM
PIK3R1	8979	NM_181523.2		2-16	Immunodeficiency 36 OMIM ?Agammaglobulinemia 7, autosomal recessive OMIM
PLCG2	9066	NM_002661.4		2-33	Autoinflammation, antibody deficiency, and immune dysregulation syndrome OMIM
PLEKHM1	29017	NM_014798.2	2-12	2-12	Osteopetrosis, autosomal recessive 6 OMIM

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PNP	7892	NM_000270.3		1-6	Immunodeficiency due to purine nucleoside phosphorylase deficiency OMIM
POLE	9177	NM_006231.3		1-49	FILS syndrome OMIM
PRF1	9360	NM_001083116.1		2-3	Aplastic anemia OMIM
PRKCD	9399	NM_006254.3		3-19	Autoimmune lymphoproliferative syndrome, type III OMIM
PRKDC	9413	NM_006904.6		1-86	Immunodeficiency 26, with or without neurologic abnormalities OMIM
PSMB8	9545	NM_148919.3		1-6	Autoinflammation, lipodystrophy, and dermatosis syndrome OMIM
PSTPIP1	9580	NM_003978.3		1-15	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne OMIM
PTPRC	9666	NM_002838.4		2-33	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive OMIM
RAB27A	9766	NM_004580.4		2-6	Griscelli syndrome, type 2 OMIM
RAC2	9802	NM_002872.4		1-6	Neutrophil immunodeficiency syndrome OMIM
RAD50	9816	NM_005732.3		1-25	Nijmegen breakage syndrome-like disorder OMIM
RAD51C	9820	NM_058216.2		1-9	Fanconi anemia, complementation group O OMIM
RAG1	9831	NM_000448.2		2	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

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RAG2	9832	NM_000536.3		2	Omenn syndrome OMIM Combined cellular and humoral immune defects with granulomas OMIM Severe combined immunodeficiency, B cell-negative OMIM
RASGRP2	9879	NM_153819.1		2-16	?Bleeding disorder, platelet-type, 18 OMIM
RBCK1	15864	NM_031229.2		1-12	Polyglucosan body myopathy 1 with or without immunodeficiency OMIM
RBM8A	9905	NM_005105.4	1-6	1-6	Thrombocytopenia-absent radius syndrome OMIM
RECQL4	9949	NM_004260.3		1-22	Rothmund-Thomson syndrome OMIM RAPADILINO syndrome OMIM Baller-Gerold syndrome OMIM PubMed
RFX5	9986	NM_000449.3		3-11	Bare lymphocyte syndrome, type II, complementation group E OMIM Bare lymphocyte syndrome, type II, complementation group C OMIM
RFXANK	9987	NM_003721.3		3-10	MHC class II deficiency, complementation group B OMIM
RFXAP	9988	NM_000538.3		1-3	Bare lymphocyte syndrome, type II, complementation group D OMIM
RHAG	10006	NM_000324.2		1-10	Overhydrated hereditary stomatocytosis OMIM Anemia, hemolytic, Rh-null, regulator type OMIM
RHOH	686	NM_004310.4		3	Kronisk epidermodysplasia verruciformis/ oeket disposisjon for HPV PubMed
RLTPR	27089	NM_001013838.1		1-38	IN PRESS ASP: 4 pasienter i 3 norske fam. med mutasjon i genet
RNASEH2A	18518	NM_006397.2		1-8	Aicardi-Goutieres syndrome 4 OMIM

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RNASEH2B	25671	NM_024570.3		1-11	Aicardi-Goutieres syndrome 2 OMIM
RNASEH2C	24116	NM_032193.3		1-4	Aicardi-Goutieres syndrome 3 OMIM
RNF168	26661	NM_152617.3		1-6	RIDDLE syndrome OMIM
RNF31	16031	NM_017999.4		1-21	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia PubMed
RORC	10260	NM_005060.3		1-11	Immunodeficiency 42 OMIM
RPL11	10301	NM_000975.3		1-6	Diamond-Blackfan anemia 7 OMIM
RPL15	10306	NM_001253384.1	2-4	2-5	?Diamond-Blackfan anemia 12 OMIM
RPL26	10327	NM_000987.3		2-4	?Diamond-Blackfan anemia 11 OMIM
RPL35A	10345	NM_000996.2		2-5	Diamond-Blackfan anemia 5 OMIM
RPL5	10360	NM_000969.3		1-8	Diamond-Blackfan anemia 6 OMIM
RPS10	10383	NM_001014.4		2-6	Diamond-Blackfan anemia 9 OMIM
RPS19	10402	NM_001022.3		2-6	Diamond-Blackfan anemia 1 OMIM
RPS24	10411	NM_033022.3		1-5	Diamond-blackfan anemia 3 OMIM
RPS26	10414	NM_001029.3		1-4	Diamond-Blackfan anemia 10 OMIM
RPS29	10419	NM_001032.4		1-3	Diamond-Blackfan anemia 13 OMIM
RPS7	10440	NM_001011.3		2-7	Diamond-Blackfan anemia 8 OMIM

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RPSA	6502	NM_002295.5		2-7	Asplenia, isolated congenital OMIM
RTEL1	15888	NM_032957.4		2-35	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3 OMIM
RUNX1	10471	NM_001754.4		2-9	Platelet disorder, familial, with associated myeloid malignancy OMIM
SAMHD1	15925	NM_015474.3		1-16	Aicardi-Goutieres syndrome 5 OMIM
SBDS	19440	NM_016038.2	1-5	1-5	Shwachman-Diamond syndrome OMIM
SERPING1	1228	NM_000062.2		2-8	Complement component 4, partial deficiency of OMIM
SH2D1A	10820	NM_002351.4		1-4	Lymphoproliferative syndrome, X-linked, 1 OMIM
SKIV2L	10898	NM_006929.4		1-28	Trichohepatoenteric syndrome 2 OMIM
SLC29A3	23096	NM_018344.5		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM
SLC35C1	20197	NM_018389.4		1-2	Congenital disorder of glycosylation, type IIc OMIM
SLC37A4	4061	NM_001164277.1		3-11	Glycogen storage disease Ib OMIM Glycogen storage disease Ic OMIM
SLC46A1	30521	NM_080669.5		1-6	Folate malabsorption, hereditary OMIM
SLX4	23845	NM_032444.2		2-15	Fanconi anemia, complementation group P OMIM
SMARCAL1	11102	NM_014140.3		3-18	Schimke immunoosseous dysplasia OMIM
SNX10	14974	NM_001199835.1		2-7	Osteopetrosis, autosomal recessive 8 OMIM
SP110	5401	NM_004509.3		2-18	Hepatic venoocclusive disease with immunodeficiency OMIM

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SPINK5	15464	NM_006846.3		1-33	Netherton syndrome OMIM Atopy OMIM
SRP72	11303	NM_006947.3	19	1-19	Bone marrow failure syndrome 1 OMIM
STAT1	11362	NM_007315.3		3-25	Immunodeficiency 31A, mycobacteriosis, autosomal dominant OMIM Immunodeficiency 31C, autosomal dominant OMIM Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive OMIM
STAT2	11363	NM_005419.3		2-24	Immunodeficiency 44 OMIM
STAT3	11364	NM_139276.2		2-24	Hyper-IgE recurrent infection syndrome OMIM Autoimmune disease, multisystem, infantile-onset OMIM
STAT5B	11367	NM_012448.3	6-9	2-19	Growth hormone insensitivity with immunodeficiency OMIM
STIM1	11386	NM_003156.3		1-12	Immunodeficiency 10 OMIM
STK4	11408	NM_006282.2		1-11	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations OMIM
STX11	11429	NM_003764.3		2	Hemophagocytic lymphohistiocytosis, familial, 4 OMIM
STXBP2	11445	NM_006949.3		1-19	Hemophagocytic lymphohistiocytosis, familial, 5 OMIM
TAP1	43	NM_000593.5		1-11	Bare lymphocyte syndrome, type I OMIM
TAP2	44	NM_000544.3		2-12	Bare lymphocyte syndrome, type I, due to TAP2 deficiency OMIM
TAPBP	11566	NM_003190.4		1-8	Bare lymphocyte syndrome, type I OMIM

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TAZ	11577	NM_000116.4		1-11	Barth syndrome OMIM
TBX1	11592	NM_080647.1		2-9	DiGeorge syndrome OMIM Velocardiofacial syndrome OMIM
TCF3	11633	NM_003200.3		2-19	Agammaglobulinemia 8, autosomal dominant OMIM
TCIRG1	11647	NM_006019.3		2-20	Osteopetrosis, autosomal recessive 1 OMIM
TCN2	11653	NM_000355.3		1-9	Transcobalamin II deficiency OMIM
TERT	11730	NM_198253.2		1-16	{Dyskeratosis congenita, autosomal recessive 4} OMIM {Dyskeratosis congenita, autosomal dominant 2} OMIM {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1} OMIM
TFRC	11763	NM_003234.2		2-19	Immunodeficiency 46 OMIM
TGFB1	11766	NM_000660.5		1-7	Camurati-Engelmann disease OMIM
THBD	11784	NM_000361.2		1	{Hemolytic uremic syndrome, atypical, susceptibility to, 6} OMIM
TICAM1	18348	NM_182919.3		2	{Herpes simplex encephalitic, susceptibility to, 6} OMIM
TINF2	11824	NM_001099274.1		1-9	Dyskeratosis congenita, autosomal dominant 3 OMIM Revesz syndrome OMIM
TLR3	11849	NM_003265.2		2-5	{Herpes simplex encephalitis, susceptibility to, 2} OMIM
TMC6	18021	NM_007267.6		2-20	Epidermodysplasia verruciformis OMIM
TMC8	20474	NM_152468.4		2-16	Epidermodysplasia verruciformis OMIM

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TMEM173	27962	NM_198282.3		3-8	STING-associated vasculopathy, infantile-onset OMIM
TNFRSF11A	11908	NM_003839.3		1-10	Osteopetrosis, autosomal recessive 7 OMIM
TNFRSF13B	18153	NM_012452.2		1-5	Immunodeficiency, common variable, 2 OMIM Immunoglobulin A deficiency 2 OMIM
TNFRSF13C	17755	NM_052945.3		1-3	Immunodeficiency, common variable, 4 OMIM
TNFRSF1A	11916	NM_001065.3		1-10	Periodic fever, familial OMIM
TNFRSF4	11918	NM_003327.3		1-7	?Immunodeficiency 16 OMIM
TNFSF11	11926	NM_003701.3		1-5	Osteopetrosis, autosomal recessive 2 OMIM
TNFSF12	11927	NM_003809.2		1-7	B celle svikt/ immunglobulinmangel/ nedsatt humoralt immunrespons PubMed
TPP2	12016	NM_003291.2		1-29	Early-onset Evans syndrome, immunodeficiency, and premature immunosenescence associated with tripeptidyl-peptidase II deficiency PubMed
TRAF3	12033	NM_003300.3		2-11	{?Herpes simplex encephalitis, susceptibility to, 3} OMIM
TRAF3IP2	1343	NM_147686.3		2-9	?Candidiasis, familial, 8 OMIM
TREX1	12269	NM_033629.4		2	Aicardi-Goutieres syndrome 1, dominant and recessive OMIM
TRNT1	17341	NM_182916.2		2-8	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay OMIM
TTC37	23639	NM_014639.3		4-43	Trichohepatoenteric syndrome 1 OMIM

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TTC7A	19750	NM_020458.3		1-20	Gastrointestinal defects and immunodeficiency syndrome OMIM
TYK2	12440	NM_003331.4		3-25	Immunodeficiency 35 OMIM
UNC119	12565	NM_005148.3		1-5	?Immunodeficiency 13 OMIM
UNC13D	23147	NM_199242.2		1-32	Hemophagocytic lymphohistiocytosis, familial, 3 OMIM
UNG	12572	NM_080911.2		1-7	Immunodeficiency with hyper IgM, type 5 OMIM
USB1	25792	NM_024598.3		1-7	Poikiloderma med neutropeni OMIM
VPS13B	2183	NM_017890.4		2-62	Cohen syndrome OMIM
VPS45	14579	NM_007259.4		1-15	Neutropenia, severe congenital, 5, autosomal recessive OMIM
WAS	12731	NM_000377.2		1-12	Neutropenia, severe congenital, X-linked OMIM Wiskott-Aldrich syndrome OMIM Thrombocytopenia, X-linked, intermittent OMIM Thrombocytopenia, X-linked OMIM
WIPF1	12736	NM_001077269.1		2-8	?Wiskott-Aldrich syndrome 2 OMIM
WRAP53	25522	NM_018081.2		1-10	Dyskeratosis congenita, autosomal recessive 3 OMIM
XIAP	592	NM_001167.3	7	2-7	Lymphoproliferative syndrome, X-linked, 2 OMIM
ZAP70	12858	NM_001079.3		3-14	Selective T-cell defect OMIM
ZBTB24	21143	NM_014797.2		2-7	Immunodeficiency-centromeric instability-facial anomalies syndrome-2 OMIM