Oslo universitetssykehus
Avdeling for medisinsk genetikk

Immunsviktsykdommer

Genpanel, versjon v04

* 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: **ACTB, C4A, C4B, CFHR1, CFHR3, CSF2RA, CYCS, FCGR1A, FCGR3A, FCGR3B, GBA, HBA1, HBA2, HBG1, HBG2, NCF1, PLEKHM1, RBM8A, RHCE, RPL15, RPS17, 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 affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ABCB7	48	NM_004299.6		1-16	Sideroblastic Anemia and Ataxia Anemia, sideroblastic, with ataxia, 301310
ABCD4	68	NM_005050.3		1-19	Methylmalonic aciduria and homocystinuria, cblJ type OMIM
ABCG5	13886	NM_022436.2		1-13	Platelet disorder Sitosterolemia and Thrombocytopenia
ABCG8	13887	NM_022437.2		1-13	Platelet disorder Sitosterolemia and Thrombocytopenia
ABL1	76	NM_005157.5		1-11	Chronic Myeloid Leukemia (CML)
ACD	25070	NM_001082486.1		1-12	?Dyskeratosis congenita, autosomal recessive 7 OMIM ?Dyskeratosis congenita, autosomal dominant 6 OMIM
ACP5	124	NM_001111035.2		4-7	Spondyloenchondrodysplasia with immune dysregulation OMIM
ACSL6	16496	NM_015256.3		1-21	Myelodysplastic syndrome Myelogenous leukemia, acute

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ACTB	132	NM_001101.4	2-6	2-6	Baraitser-Winter syndrome 1 with macrothrombocytopenia Platelet disorder Baraitser-Winter syndrome 1, 243310 Phagocytic disorder Actin beta deficiency (ACTB) neutrophil dysfunction Poor neutrophil chemotaxis, oxidative burst and actin remodeling. Thrombocytopenia Mental retardation, short stature Congenital defects of phagocyte number or function
ACTN1	163	NM_001130004.1		1-22	Macrothrombocytopenia
ADA	186	NM_000022.3		1-12	Severe combined immunodeficiency due to ADA deficiency OMIM Adenosine deaminase deficiency, partial OMIM
ADAM17	195	NM_003183.6		1-19	Inflammatory skin and bowel disease, neonatal, 1 IBD-1 inflammatory skin Inflammatory skin and bowel disease, neonatal 1, 614328 ADAM17 deficiency Early onset diarrhea and skin lesions Autoinflammatory Disorders
ADAMTS13	1366	NM_139025.4		1-29	Familial Thrombotic Thrombocytopenia Purpura Thrombotic thrombocytopenic purpura, familial, 274150 Congenital Thrombotic Thrombocytopenic Purpura Schulman-Upshaw Syndrome Familial thrombotic thrombocytopenic purpura TTP Thrombotic disorder Thrombotic thrombocytopenic purpura, familial
ADAR	225	NM_001111.5		1-15	Dyschromatosis symmetrica hereditaria OMIM Aicardi-Goutieres syndrome 6 OMIM
AICDA	13203	NM_020661.3		1-5	Immunodeficiency with hyper-IgM, type 2 OMIM
AIRE	360	NM_000383.3		1-14	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia OMIM
AK1	361	NM_000476.2		2-7	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	362	NM_001625.3		1-6	Reticular dysgenesis OMIM
ALAS2	397	NM_000032.5		2-11	Anemia, sideroblastic, 1 300751

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ALDOA	414	NM_000034.3		7-14	Enzyme Disorder Glycogen storage disease Glycogen storage disease XII, 611881 Aldolase A deficiency Glycogen storage disease due to aldolase A deficiency
AMN	14604	NM_030943.3		1-12	Megaloblastic anemia-1, Norwegian type, 261100
ANK1	492	NM_000037.3		1-42	RBC membrane abnormality Spherocytosis, type 1,182900
ANKRD26	29186	NM_014915.2		1-34	Thrombocytopenia 2, 188000 Congenital amegkaryocytic thrombocytopenia Thrombocytopenia 2 610855 610855 (OMIN gene description ID) not submitted (OMIM phenotype description ID)
ANKRD34A	27639	NM_001039888.3		4	Thrombocytopenia Absent-Radius Syndrome
ANKRD35	26323	NM_144698.4		1-13	Thrombocytopenia Absent-Radius Syndrome
AP1S3	18971	NM_001039569.1		1-5	Pustular psoriasis, 616106 Autoinflammatory Disorders
AP3B1	566	NM_003664.4		1-27	Hermansky-Pudlak syndrome 2 OMIM
AP3D1	568	NM_001261826.3		1-32	?Hermansky-Pudlak syndrome 10 Platelet disorder Hermansky-Pudlak syndrome and MR ?Hermansky-Pudlak syndrome 10, 617050 HSP10 Immunodeficient HPS Hermansky-Pudlak syndrome with neutropenia Hermansky-Pudlak syndrome albinism neutropenia neuordevelopmental delay seizures Oculocutaneous albinism, severe neutropenia, recurrent infections, seizures, hearing loss and neurodevelopmental delay Diseases of Immune Dysregulation
APOL1	618	NM_003661.3	5-6	2-6	Trypanosomiasis, susceptibility to Trypanosomias Trypanosomiasis Defects in Intrinsic and Innate Immunity
ARHGAP26	17073	NM_001135608.2		1-23	Myelodysplastic syndrome (MDS), Paediatric Leukemia, juvenile myelomonocytic, somatic 607785

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ARPC1B	704	NM_005720.4		2-10	Platelet disorder Thrombocytopenia and Immune Deficiency Thrombocytopenia & Immune Deficiency Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718 inflammatory predisposition Immunodeficiency with thrombocytopenia Mild thrombocytopenia with normal sized platelets, recurrent invasive infections, colitis, vasculitis, autoantibodies (ANA, ANCA), eosinophilia, defective Arp2/3, filament branching Combined immunodeficiencies with associated or syndromic features
ASXL1	18318	NM_015338.5		1-12	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286 juvenile myelomonocytic leukaemia aquired aplastic anaemia
ATM	795	NM_000051.3		2-63	Ataxia-telangiectasia OMIM
ATP6AP1	868	NM_001183.5		1-10	Immunodeficiency 47, 300972 Hepatopathy, leukopenia, low copper Predominantly Antibody Deficiencies Immunodeficiency and hepatopathy with or without neurologic features
ATP7B	870	NM_000053.3		1-21	Wilson disease OMIM
ATR	882	NM_001184.3		1-47	Seckel syndrome 1 OMIM
ATRX	886	NM_000489.5		1-35	Myelodysplastic syndrome (MDS), Adult Alpha-thalassemia myelodysplasia syndrome, somatic 300448 Alpha-thalassemia/mental retardation syndrome 301040 XLD
B2M	914	NM_004048.3		1-3	Immunodeficiency 43 OMIM
BAAT	932	NM_001701.3		2-4	Hypercholanemia, familial, 607748 Hypercholanemia
BACH2	14078	NM_021813.3		8-9,6-7	Lymphocytic colitis, sinopulmonary infections Diseases of Immune Dysregulation infantile onset enterocolitis hypogammaglobulinaemia BACH2-related immunodeficiency and autoimmunity (BRIDA)
BCL10	989	NM_003921.5		1-3	?Immunodeficiency 37 OMIM
BCL11B	13222	NM_138576.3		1-4	?Immunodeficiency 49, 617237 Congenital abnormalities, neonatal teeth, dysmorphic facies, absent corpus callosum, neurocognitive deficits Immunodeficiencies affecting cellular and humoral immunity leaky SCID

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
BCOR	20893	NM_017745.5		2-15	Acute myeloid leukaemia (AML) aquired aplastic anaemia
BCORL1	25657	NM_021946.4		1-12	Acute myeloid leukaemia (AML) aquired aplastic anaemia
BLM	1058	NM_000057.3		2-22	Bloom syndrome OMIM
BLNK	14211	NM_013314.3		1-17	Agammaglobulinemia 4 OMIM
BLOC1S6	8549	NM_012388.3		1-5	Hermansky-pudlak syndrome 9 OMIM
BRAF	1097	NM_004333.5	18	1-18	Rasopathies Leukaemia Lymphoma Hairy cell leukemia (HCL)
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
C19orf40	28467	NM_152266.4		2-5	EBV infection-driven lymphoproliferative disease Diseases of Immune Dysregulation
C1QA	1241	NM_015991.3		2-3	C1q deficiency OMIM
C1QB	1242	NM_000491.4		2-3	C1q deficiency OMIM
C1QC	1245	NM_172369.4		2-3	C1q deficiency OMIM
C1S	1247	NM_201442.3		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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
C4A	1323	NM_007293.2	1-41	1-41	C4a deficiency, 614380 Complement component 4 deficiency Immunodeficiency due to a classical component pathway complement deficiency SLE predisposition infections with encapsulated organisms SLE, infections with encapsulated organisms , partial deficiency is common (either C4A or C4B) and appears to have a modest effect on host defense Complement Deficiencies
C4B	1324	NM_001002029.3	1-41	1-41	C4B deficiency, 614379 SLE predisposition SLE, infections with encapsulated organisms , partial deficiency is common (either C4A or C4B) and appears to have a modest effect on host defense Complement Deficiencies
C5	1331	NM_001735.2		1-41	C5 deficiency OMIM
C6	1339	NM_000065.3		2-18	C6 deficiency OMIM
C6orf25	13937	NM_138272.2		1-6	?Thrombocytopenia, anemia, and myelofibrosis 617441 Platelet disorder Thrombocytopenia, anemia, and myelofibrosis
C7	1346	NM_000587.3		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
C8G	1354	NM_000606.3		1-7	Complement factor 8 defect Complement component 8 deficiency Disseminated neisserial infections Complement Deficiencies
C9	1358	NM_001737.4		1-11	C9 deficiency OMIM
CALR	1455	NM_004343.3		1-9	Myelofibrosis Essential thrombocythemia (ET) Myelofibrosis, somatic, 254450 Thrombocythemia, somatic,187950
CARD11	16393	NM_032415.5		2-25	Immunodeficiency 11 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CARD14	16446	NM_024110.4		2-21	CARD14 mediated psoriasis Psoriasis 2, 602723 Pityriasis rubra pilaris,173200 Other autoinflammatory diseases with known genetic defect Psoriasis Autoinflammatory Disorders immune dysregulation
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
CBL	1541	NM_005188.3		1-16	Myelodysplastic syndrome (MDS), Paediatric Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CBLB	1542	NM_170662.5		2-19	Acute myeloid leukaemia (AML)
CBLC	15961	NM_012116.4		1-10	Chronic Myeloid Leukemia (CML)
CCBE1	29426	NM_133459.4		1-11	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 Lymphangiectasia and lymphedema with facial abnormalities and other dysmorphic features Combined immunodeficiencies with associated or syndromic features
CD19	1633	NM_001770.5		1-14	Immunodeficiency, common variable, 3 OMIM
CD247	1677	NM_198053.2		1-8	?Immunodeficiency 25 OMIM
CD27	11922	NM_001242.4		1-6	Lymphoproliferative syndrome 2 OMIM
CD320	16692	NM_016579.3		1-5	Methylmalonic aciduria, transient, due to transcobalamin receptor defect OMIM
CD36	1663	NM_001001547.2		3-14	[Macrothrombocytopenia] (1)
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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CD4	1678	NM_000616.4		2-10	Selective CD4 cell deficiency
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
CD46	6953	NM_002389.4	2-5	1-13	Hemolytic uremic syndrome, atypical, susceptibility to, 2, 612922 Membrane Cofactor Protein (CD46) deficiency atypical HUS Atypical hemolytic-uremic syndrome, infections, preeclampsia Complement Deficiencies
CD55	2665	NM_000574.4		1-10	Decay-accelerating factor for complement deficiency (DAF CD55) protein-losing enteropathy hypogammaglobulinaemia angiopathic thrombosis primary intestinal lymphangiectasia Protein losing enteropathy, thrombosis Complement Deficiencies
CD59	1689	NM_203330.2		4-6	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 CD59 antigen P18-20 deficiency (CD59) Membrane Attack Complex Inhibitor (CD59) deficiency Primary CD59 deficiency paroxysmal nocturnal haemoglobinuria chronic hemolysis childhood relapsing immune-mediated polyneuropathy Hemolytic anemia, polyneuropathy Complement Deficiencies
CD70	11937	NM_001252.4		1-3	Combined immunodeficiency CD70-deficiency EBV-related malignancy EBV susceptibility, Hodgkin lymphoma Diseases of Immune Dysregulation
CD79A	1698	NM_001783.3		1-5	Agammaglobulinemia 3 OMIM
CD79B	1699	NM_000626.3		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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CDAN1	1713	NM_138477.4		1-28	Dyserythropoietic anemia, congenital, type Ia OMIM
CDCA7	14628	NM_031942.4		1-10	Immunodeficiency-centromeric instability-facial anomalies syndrome ICF Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 ICF3 immunodeficiency, centromeric instability, facial anomalies syndrome type 3 recurrent respiratory infections hypogammaglobulinaemia enteropathy Facial dysmorphic features, macroglossia, bacterial/opportunistic infections, malabsorption, cytopenias, malignancies, multiradial configurations of chromosomes 1, 9, 16 Combined immunodeficiencies with associated or syndromic features
CDKN2A	1787	NM_000077.4		1-3	Acute lymphoblastic leukemia (ALL)
CEBPA	1833	NM_004364.4		1	Familial MDS (Myelodysplastic syndromes) acute myeloid leukemia (AML) 601626 (OMIM phenotype description ID) 116897 (OMIN gene description ID) 116897 / 601626 Leukemia, acute myeloid, somatic 601626 Leukemia, acute myeloid, somatic
CEBPE	1836	NM_001805.3		1-2	Specific granule deficiency OMIM
CECR1	1839	NM_001282228.1		2-10	DBA Diamond-Blackfan Anemia Polyarteritis nodosa Polyarteritis nodosa, childhood-onset, 615688 ADA2 deficiency Deficiency of ADA2 (DADA2) Other autoinflammatory diseases with known genetic defect Fever with early onset stroke combined immunodeficiency Evans' syndrome Polyarteritis nodosa, childhood-onset, early-onset recurrent ischemic stroke and fever Autoinflammatory Disorders
CFB	1037	NM_001710.5		1-18	?Complement factor B deficiency OMIM
CFD	2771	NM_001928.3		1-5	Complement factor D deficiency OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CFH	4883	NM_000186.3	8-10, 20-22	1-22	Complement factor H deficiency OMIM
CFHR1	4888	NM_002113.2	1-6	1-6	Age related macular degeneration Atypical hemolytic uremic syndrome susceptibility Older onset atypical hemolytic-uremic syndrome, disseminated neisserial infections Complement Deficiencies
CFHR2	4890	NM_005666.4	1-3	1-5	Age related macular degeneration Atypical hemolytic uremic syndrome susceptibility Older onset atypical hemolytic-uremic syndrome, disseminated neisserial infections Complement Deficiencies
CFHR3	16980	NM_021023.5	1-6	1-6	Age related macular degeneration Atypical hemolytic uremic syndrome susceptibility Older onset atypical hemolytic-uremic syndrome, disseminated neisserial infections Complement Deficiencies
CFHR4	16979	NM_001201550.3	7-10	1-10	Age related macular degeneration Atypical hemolytic uremic syndrome susceptibility Older onset atypical hemolytic-uremic syndrome, disseminated neisserial infections Complement Deficiencies
CFHR5	24668	NM_030787.3		1-10	Nephropathy due to CFHR5 deficiency OMIM
CFI	5394	NM_000204.4		1-13	Complement factor I deficiency OMIM
CFP	8864	NM_002621.2		2-10	PROPERDIN DEFICIENCY, X-LINKED OMIM
CFTR	1884	NM_000492.3		1-27	Cystic fibrosis, 219700 Respiratory infections, pancreatic insufficiency, elevated sweat chloride Congenital defects of phagocyte number or function
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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CLPB	30664	NM_030813.5		1-17	3-methylglutaconic aciduria, type VII 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 3-methylglutaconic aciduria, type 7 Recurrent or severe infection Neurocognitive developmental aberrations, microcephaly, hypoglycemia, hypotonia, ataxia, seizures, cataracts, IUGR Congenital defects of phagocyte number or function
CNBP	13164	NM_003418.4		2-5	Steinert- myotonica dystrophia
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
COX4I2	16232	NM_032609.2		2-5	Exocrine Pancreatic Insufficiency, Dyserythropoietic Anemia, andCalvarial Hyperostosis Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
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_025099.6		1-23	Cerebroretinal microangiopathy with calcifications and cysts OMIM
CTLA4	2505	NM_005214.5		1-4	Autoimmune lymphoproliferative syndrome, type V OMIM
CTPS1	2519	NM_001905.4		2-18	Immunodeficiency 24 OMIM
CTSC	2528	NM_001814.5		1-7	Haim-Munk syndrome OMIM Periodontitis 1, juvenile OMIM
CUBN	2548	NM_001081.3	41-50, 61-67	1-67	Megaloblastic anemia-1, Finnish type, 261100 Megaloblastic Anemia
CUX1	2557	NM_001202543.1	3	1-24	Acute myeloid leukaemia (AML)
CXCR4	2561	NM_003467.2		1-2	WHIM syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CYB5R3	2873	NM_000398.6		1-9	Methemoglobinemia, type I OMIM Methemoglobinemia, type II 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
CYCS	19986	NM_018947.5	2-3	2-3	Thrombocytopenia 4, 612004 Thrombocytopenia 4
DCLRE1B	17641	NM_022836.4		1-4	Hoyeraal-Hreidarsson syndrome Intrauterine growth retardation, microcephaly, nail dystrophy, sparse scalp hair and eyelashes, hyperpigmentation of skin, palmar hyperkeratosis, premalignant oral leukoplakia, pancytopenia, myelodysplasia, +/- recurrent infections. A severe phenotype with developmental delay and cerebellar hypoplasia known as Hoyeraal-Hreidarsson Syndrome (HHS) may occur in some DKC patients Combined immunodeficiencies with associated or syndromic features
DCLRE1C	17642	NM_001033855.2	4-9	1-14	Severe combined immunodeficiency, Athabascan type OMIM Omenn syndrome OMIM
DDX41	18674	NM_016222.3		1-17	616871 {Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to} 616871 (OMIM phenotype description ID)
DDX58	19102	NM_014314.3		1-18	Associations with humoral and cellular immunity to measles vaccine
DGKE	2852	NM_003647.2		2-12	Nephrotic syndrome, type 7 OMIM
DHFR	2861	NM_000791.3	6	1-6	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	2890	NM_001363.4		1-15	Dyskeratosis congenita, X-linked OMIM
DNAJC21	27030	NM_001012339.3		1-12	Pancytopaenia Bone Marrow Failure Bone marrow failure syndrome 3, 617052 Bone marrow failure syndrome 3, 617052 Shwachman-Diamond syndrome-like Metaphyseal changes, short stature, developmental delay, pancreatic dysfunction, bone marrow failure Congenital defects of phagocyte number or function
DNASE1L3	2959	NM_004944.3		1-8	Systemic lupus erythematosus 16, 614420 familial early-onset SLE Systemic lupus erythematosus, lupus nephritis, hypocomplementemic urticarial vasculitis Diseases of Immune Dysregulation

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
DNASE2	2960	NM_001375.2		1-6	Glomerulonephritis, arthropathy, vasculitis Autoinflammatory Disorders multisystem autoinflammatory syndrome severe neonatal anemia membranoproliferative glomerulonephritis liver fibrosis deforming arthropathy SLE
DNMT3A	2978	NM_175629.2		2-23	Myelodysplastic syndrome (MDS), Paediatric acute myeloid leukaemia aquired aplastic anaemia
DNMT3B	2979	NM_006892.3		2-23	Immunodeficiency-centromeric instability-facial anomalies syndrome 1 OMIM
DOCK2	2988	NM_004946.3		1-52	Immunodeficiency 40 OMIM
DOCK8	19191	NM_203447.3		1-48	Hyper-IgE recurrent infection syndrome, autosomal recessive OMIM
EFTUD1	25789	NM_024580.5	2-15	2-20	Shwachman-Diamond syndrome 2 OMIM
ELANE	3309	NM_001972.3		1-5	Neutropenia, cyclic OMIM Neutropenia, severe congenital 1, autosomal dominant OMIM
ELF4	3319	NM_001421.3		2-9	X-linked hypogammaglobulinemia with isolated growth hormone deficiency
ENO1	3350	NM_001428.5		2-12	Enzyme Disorder Enolase deficiency
EPB41	3377	NM_004437.3		4-18	RBC membrane abnormality Elliptocytosis Elliptocytosis-1,611804 Hereditary elliptocytosis
EPB42	3381	NM_000119.3		1-13	RBC membrane abnormality Elliptocytosis Spherocytosis, type 5, 612690 Hereditary spherocytosis type 5 Minkowski-Chauffard disease Spherocytosis, Recessive EPB42-related hereditary spherocytosis
EPCAM	11529	NM_002354.2		1-9	Diarrhea 5, with tufting enteropathy, congenital
EPG5	29331	NM_020964.3		1-44	Vici syndrome OMIM
EPHX1	3401	NM_000120.3		2-9	?Fetal hydantoin syndromeDiphenylhydantoin toxicityHypercholanemia, familial, 607748{Preeclampsia, susceptibility to}, 189800

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
EPO	3415	NM_000799.3		1-5	DBA Diamond-Blackfan anemia-like (AR), 617911 Erythrocytosis, familial 5 (AD), 617907
ERCC2	3434	NM_000400.3		1-23	Trichothiodystrophy 1, photosensitive OMIM
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
ETV6	3495	NM_001987.4		1-8	Thrombocytopenia, red cell macrocytosis and predisposition to lymphoblastic leukaemia 600618 Thrombocytopenia 5 601626 Leukemia, acute myeloid, somatic 601626 Leukemia, acute myeloid, somatic
EXTL3	3518	NM_001440.3		3-7	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 EXTL3 deficiency Platyspondyly, kyphosis, variable skeletal dysplasias, developmental delay Combined immunodeficiencies with associated or syndromic features
EZH2	3527	NM_004456.4		2-20	Myelodysplastic syndrome (MDS), Paediatric
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
FAM105B	25118	NM_138348.5		1-7	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 Fever, diarrhea , dermatitis Autoinflammatory Disorders
FANCA	3582	NM_000135.3		1-43	Fanconi anemia, complementation group A OMIM
FANCB	3583	NM_001018113.2		3-10	Fanconi anemia, complementation group B OMIM
FANCC	3584	NM_000136.2		2-15	Fanconi anemia, complementation group C OMIM
FANCD2	3585	NM_033084.4	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

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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
FANCM	23168	NM_020937.3		1-23	Fanconi Anemia Fanconi anemia, complementation group M, 614087
FAS	11920	NM_000043.5		1-9	{Autoimmune lymphoproliferative syndrome} OMIM Autoimmune lymphoproliferative syndrome, type IA OMIM
FASLG	11936	NM_000639.2		1-4	Autoimmune lymphoproliferative syndrome, type IB OMIM
FAT4	23109	NM_024582.4		1-17	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Lymphangiectasia and lymphedema with facial abnormalities and other dysmorphic features Combined immunodeficiencies with associated or syndromic features
FBXW7	16712	NM_033632.3		2-12	Acute lymphoblastic leukemia (ALL)
FCGR1A	3613	NM_000566.3	1-6	1-6	Fc receptor deficiencies
FCGR2A	3616	NM_021642.3	4-7	1-7	Fc receptor deficiencies
FCGR2B	3618	NM_004001.4	1-6	1-8	Fc receptor deficiencies
FCGR3A	3619	NM_000569.7	1-5	1-5	Immunodeficiency 20 OMIM
FCGR3B	3620	NM_000570.4	2-6	2-6	Neutropenia,alloimmuneneonatal Neutropenia,alloimmuneneonatal Neutropenia, alloimmune neonatal Fc receptor deficiencies Neutropenia, autoimmune neonatal Neutropenia, autoimmune neonatal
FCGRT	3621	NM_001136019.2		2-7	Fc receptor deficiencies
FCN3	3625	NM_003665.3		1-8	Immunodeficiency due to ficolin 3 deficiency OMIM
FECH	3647	NM_000140.3		1-11	Protoporphyria, erythropoietic, 1 OMIM

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FERMT3	23151	NM_031471.5		2-15	Leukocyte adhesion deficiency, type III OMIM
FLI1	3749	NM_002017.4		1-9	Paris-Trousseau thrombocytopenia and Jacobson syndrome (BIALLELIC, autosomal or pseudoautosomal) Bleeding disorder, platelet-type, 21 617443 BDPLT21 BDPLT21, Storage Pool Disorder and Bleeding (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown) BDPLT21, Paris Trousseau like (BIALLELIC, autosomal or pseudoautosomal) Bleeding disorder, platelet-type, 21
FLT3	3765	NM_004119.2		1-24	Myelodysplastic syndrome (MDS), Paediatric
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
FPR1	3826	NM_002029.3		2	Periodontitis Localized juvenile peridontitis Periodontitis only Congenital defects of phagocyte number or function
FPR2	3827	NM_001005738.1		2	Localized juvenile peridontitis
FPR3	3828	NM_002030.4		2	Localized juvenile peridontitis
FYB	4036	NM_001243093.1		1-19	Thrombocytopenia 3, 273900 Platelet disorder Microthrombocytopenia 3
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
GAD1	4092	NM_000817.2		2-17	?Cerebral palsy, spastic quadriplegic, 1, 603513
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

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GBA	4177	NM_001005741.2	2-12	2-12	Gaucher disease Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II,230800 Gaucher disease, type III, 230800 Gaucher disease, type IIIC, 231005
GCLC	4311	NM_001498.4		1-16	Enzyme Disorder Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 Glutamate-cysteine ligase deficiency
GFI1	4237	NM_005263.4		2-7	Neutropenia, nonimmune chronic idiopathic, of adults OMIM Neutropenia, severe congenital 2, autosomal dominant OMIM
GIF	4268	NM_005142.2		1-9	Intrinsic factor deficiency OMIM
GINS1	28980	NM_021067.4		1-7	GINS1 deficiency intrauterine growth retardation chronic neutropenia NK cell deficiency Immunodeficiency 55, 617827 Neutropenia, IUGR, NK cells very low Combined immunodeficiencies with associated or syndromic features
GLRX5	20134	NM_016417.2		1-2	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GNAS	4392	NM_000516.5		1-13	Acute myeloid leukaemia (AML)
GP1BA	4439	NM_000173.6		2	Bernard-Soulier syndrome, type A1 (recessive) 231200 Platelet-type von Willebrand disease Bernard-Soulier syndrome Bernard-Soulier syndrome (BIALLELIC, autosomal or pseudoautosomal) Platelet-type von Willebrand disease (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown)
GP1BB	4440	NM_000407.4		1-2	Bernard-Soulier syndrome, type B (BIALLELIC, autosomal or pseudoautosomal) Giant platelet disorder, isolated (AR) 231200 Bernard-Soulier syndrome Macrothrombocytopenia (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown)
GP9	4444	NM_000174.4		3	Bernard-Soulier syndrome, type C 231200 Bernard-Soulier syndrome
GPI	4458	NM_000175.5		1-18	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470

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GPX1	4553	NM_000581.3		1-2	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GSR	4623	NM_000637.4		1-13	Enzyme Disorder Hemolytic anemia due to glutathione reductase deficiency
GSS	4624	NM_000178.4		2-13	Enzyme Disorder Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GTF2H5	21157	NM_207118.2		2-3	Trichothiodystrophy 3, photosensitive OMIM
GUCY2C	4688	NM_004963.3		1-27	Diarrhea 6, 614616 meconium ileus, 614665
HAX1	16915	NM_006118.3		1-7	Neutropenia, severe congenital 3, autosomal recessive OMIM
HBA1	4823	NM_000558.4	1-3	1-3	Globin Disorder Erythremias, alpha- Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemias, alpha- Thalassemias, alpha-, 604131
HBA2	4824	NM_000517.4	1-3	1-3	Globin Disorder Erythrocytosis Heinz body anemia,140700 Hemoglobin H disease, nondeletional, 613978 Hypochromic microcytic anemia Thalassemia, alpha-, 60413
HBB	4827	NM_000518.4		1-3	Globin Disorder Delta-beta thalassemia (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown), 141749 Erythremias, beta- Heinz body anemias, beta- (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown), 140700 Hereditary persistence of fetal hemoglobin,(MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown),141749 Methemoglobinemias, beta- Sickle cell anemia (BIALLELIC, autosomal or pseudoautosomal),603903 Thalassemia-beta, dominant inclusion-body, 603902 Thalassemias, beta-,(BIALLELIC, autosomal or pseudoautosomal), 613985
HBD	4829	NM_000519.3		1-3	Thalassemia,delta ThalassemiaduetoHbLepore
HBG1	4831	NM_000559.2	1-3	1-3	Globin Disorder Fetal hemoglobin quantitative trait locus 1, 141749

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HBG2	4832	NM_000184.2	1-3	1-3	Globin Disorder Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1,141749
HELLS	4861	NM_018063.4		1-22	Immunodeficiency-centromeric instability-facial anomalies syndrome ICF Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 ICF4 Facial dysmorphic features, macroglossia, bacterial/opportunistic infections, malabsorption, cytopenias, malignancies, multiradial configurations of chromosomes 1, 9, 16 Combined immunodeficiencies with associated or syndromic features
HFE	4886	NM_000410.3		1-6	Hemochromatosis OMIM {Porphyria cutanea tarda, susceptibility to} OMIM {Porphyria variegata, susceptibility to} OMIM
HK1	4922	NM_000188.2		1-18	Enzyme Disorder Hemolytic anemia due to hexokinase deficiency, 235700
HMOX1	5013	NM_002133.2		1-5	Hemolysis, nephritis, inflammation Defects in Intrinsic and Innate Immunity amyloidosis
HOXA11	5101	NM_005523.5		1-2	Congenital amegkaryocytic thrombocytopenia Radioulnar synostosis with amegakaryocytic thrombocytopenia, 605432 Amegakaryocytic thrombocytopenia with radioulnar synostosis
HPS1	5163	NM_000195.4	4-6	3-20	Hermansky-Pudlak syndrome 1 OMIM
HPS4	15844	NM_022081.5		2-14	Hermansky-Pudlak syndrome 4 OMIM
HPS6	18817	NM_024747.5		1	Hermansky-Pudlak syndrome 6 OMIM
HRAS	5173	NM_005343.3		2-5	Myelodysplastic syndrome (MDS), Adult
HSPA9	5244	NM_004134.6		1-17	Anemia, sideroblastic, 4 OMIM
HTRA2	14348	NM_013247.4		1-8	3-methylglutaconic aciduria, type VIII, 617248 early onset neurological syndrome neutropenia
HYOU1	16931	NM_006389.4		2-26	Hypoglycemia, inflammatory complications Congenital defects of phagocyte number or function
ICOS	5351	NM_012092.3		1-5	Immunodeficiency, common variable, 1 OMIM

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IDH1	5382	NM_005896.3		3-10	Acute myeloid leukaemia (AML)
IDH2	5383	NM_002168.3		1-11	Acute myeloid leukaemia (AML)
IFIH1	18873	NM_022168.3		1-16	Singleton-Merten syndrome 1 OMIM Aicardi-Goutieres syndrome 7 OMIM
IFNAR2	5433	NM_207585.2		2-9	?Immunodeficiency 45, 616669 Severe viral infections (disseminated vaccine-strain measles, HHV6) Defects in Intrinsic and Innate Immunity
IFNG	5438	NM_000619.2		1-4	Aplastic Anemia
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
IKBKG	5961	NM_001099857.2	3-10	2-10	Immunodeficiency 33, 300636 Ectodermal dysplasia, hypohidrotic, with immune deficiency 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 Invasive pneumococcal disease, recurrent isolated, 2,300640 Defects of TLR/NFkappa-B signalling Anhidrotic ectodermal dysplasia (in some), various infections (bacteria, mycobacteria, viruses and fungi), colitis, conical teeth, variable defects of skin, hair and teeth, monocyte dysfunction Combined immunodeficiencies with associated or syndromic features
IKZF1	13176	NM_006060.6		2-8	Immunodeficiency, common variable, 13 OMIM
IL10	5962	NM_000572.3		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

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IL12A	5969	NM_000882.3		1-7	
IL12B	5970	NM_002187.2		2-7	Immunodeficiency 29, mycobacteriosis OMIM
IL12RB1	5971	NM_005535.2		1-17	Immunodeficiency 30 OMIM
IL12RB2	5972	NM_001559.2		2-16	
IL17A	5981	NM_002190.3		1-3	Immunodeficiency 5 Arthritis
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_153461.3		1-19	Candidiasis, familial, 9 OMIM
IL18	5986	NM_001562.3	3	2-6	Defects with susceptibility to mycobacterial infection (MSMD)
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
IL22	14900	NM_020525.4		1-5	AutoAb Chronic Mucocutaneous Candidiasis
IL23A	15488	NM_016584.2		1-4	Defects with susceptibility to mycobacterial infection (MSMD)
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_002185.4		1-8	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type OMIM
INO80	26956	NM_017553.3		2-36	INO80 deficiency, HIGM severe bacterial infections Severe bacterial infections Predominantly Antibody Deficiencies

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IRAK1	6112	NM_001569.3		1-14	Bacterial infections, X-linked MECP2 deficiency-related syndrome due to a large de novo Xq28 chromosomal deletion encompassing both MECP2 and IRAK1 Defects in Intrinsic and Innate Immunity
IRAK4	17967	NM_016123.3		2-12	Invasive pneumococcal disease, recurrent isolated, 1 OMIM IRAK4 deficiency OMIM
IRF1	6116	NM_002198.2		2-10	Myelodysplastic syndrome, preleukemic Myelogenous leukemia, acute Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980
IRF2BP2	21729	NM_182972.2		1-2	Recurrent infections, possible autoimmunity and inflammatory disease Predominantly Antibody Deficiencies CVID
IRF3	6118	NM_001571.5		2-8	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 Herpes simplex virus 1 encephalitis Defects in Intrinsic and Innate Immunity
IRF7	6122	NM_004031.3		2-10	?Immunodeficiency 39 OMIM
IRF8	5358	NM_002163.3		2-9	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive OMIM Immunodeficiency 32A, mycobacteriosis, autosomal dominant OMIM
ISG15	4053	NM_005101.4		1-2	Immunodeficiency 38 OMIM
ITCH	13890	NM_031483.6		3-25	Autoimmune disease, multisystem, with facial dysmorphism OMIM
ITGA10	6135	NM_003637.4		1-30	Thrombocytopenia Absent-Radius Syndrome
ITGA2	6137	NM_002203.3		1-30	?Glycoprotein Ia deficiency OMIM
ITGA2B	6138	NM_000419.4		1-30	BAK platelet antigen Glanzmann thrombasthenia, 273800 Glanzmann thrombasthenia (BIALLELIC, autosomal or pseudoautosomal) 273800 Bleeding disorder, platelet-type, 16, autosomal dominant (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown) 187800 BDPLT16 Congenital macrothrombocytopenia
ITGAM	6149	NM_000632.3		1-30	Systemic lupus erythematous

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ITGB2	6155	NM_000211.4		2-16	Leukocyte adhesion deficiency OMIM
ITGB3	6156	NM_000212.2		1-15	PL(A) platelet antigen Glanzmann thrombasthenia, 273800 Glanzmann thrombasthenia (BIALLELIC, autosomal or pseudoautosomal) 273800 Bleeding disorder, platelet-type, 16, autosomal dominant (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown) 187800 BDPLT16 Congenital macrothrombocytopenia
ITK	6171	NM_005546.3		1-17	Lymphoproliferative syndrome 1 OMIM
JAGN1	26926	NM_032492.4		1-2	Neutropenia, severe congenital, 6, autosomal recessive OMIM
JAK1	6190	NM_002227.3		2-25	HSM, eosinophilia, eosinophilic enteritis, thyroid disease, poor growth, viral infections Diseases of Immune Dysregulation Susceptibility to mycobacteria and viruses, urothelial carcinoma Defects in Intrinsic and Innate Immunity Hypereosinophilic syndrome
JAK2	6192	NM_004972.3		3-25	Myeloproliferative neoplasms (MPN) Acute myeloid leukaemia (AML)
JAK3	6193	NM_000215.3		2-24	SCID, autosomal recessive, T-negative/B-positive type OMIM
KCNN4	6293	NM_002250.2		1-8	Dehydrated hereditary stomatocytosis 2 OMIM
KDM6A	12637	NM_021140.3		1-29	Acute myeloid leukaemia (AML) Kabuki Syndrome 2 due to KDM6A deficiency Combined immunodeficiencies with associated or syndromic features
KIF23	6392	NM_138555.4		1-23	Enzyme Disorder Congenital dyserythropoietic anemia type III CDA III Congenital dyserythropoietic anemia (CDA)
KIT	6342	NM_000222.2		1-21	Acute myeloid leukaemia (AML)
KLF1	6345	NM_006563.4		1-3	Congenital Dyserythropoietic Anemia Dyserythropoietic anemia, congenital, type IV, 613673
KMT2A	7132	NM_001197104.1		1-36	Acute myeloid leukaemia (AML) Unclassified antibody deficiency Wiedemann-Steiner syndrome with Congenital immunodeficiency

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KMT2D	7133	NM_003482.3		1-54	Kabuki syndrome 1 OMIM
KRAS	6407	NM_004985.4	5	2-5	Myelodysplastic syndrome (MDS), Paediatric RAS associated lymphoproliferative disease, 614470 RALD
LAMTOR2	29796	NM_014017.3		1-4	Immunodeficiency due to defect in MAPBP-interacting protein OMIM
LAT	18874	NM_001014987.1		1-11	Immunodeficiency 52, 617514 Immunodeficiency 52, 617514 Immunodeficiency 52, 617514 Adenopathy, splenomegaly, recurrent infections, autoimmunity Immunodeficiencies affecting cellular and humoral immunity
LCAT	6522	NM_000229.1		1-6	Norum disease OMIM
LCK	6524	NM_001042771.2		2-13	?Immunodeficiency 22 OMIM
LIG1	6598	NM_000234.2		2-28	DNA ligase I deficiency DNA-ligase 1 ATP-dependent deficiency (LIG1) Recurrent respiratory infections, growth retardation, sun sensitivity, lymphoma, radiation sensitivity Combined immunodeficiencies with associated or syndromic features
LIG4	6601	NM_002312.3		2	LIG4 syndrome OMIM
LIX1L	28715	NM_153713.2		1-6	Thrombocytopenia Absent-Radius Syndrome
LMBRD1	23038	NM_018368.3		1-16	Methylmalonic aciduria and homocystinuria, cblF type 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_006785.4		1-17	Immunodeficiency 12 OMIM

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MASP1	6901	NM_139125.3		1-11	Mannan-binding lectin serine protease (MASP) deficiency
MASP2	6902	NM_006610.3		1-11	MASP2 deficiency OMIM
MASTL	19042	NM_032844.4		1-12	Thrombocytopenia severe aplastic anemia
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
MECOM	3498	NM_001105078.3		3-16	transcription factor and gives the same pathology (thrombocytopenia with bone defects) as HOXA11 Radioulnar synostosis with amegakaryocytic thrombocytopenia 2
MEFV	6998	NM_000243.2		1-10	Familial Mediterranean fever, AR OMIM Familial Mediterranean fever, AD OMIM
MKL1	14334	NM_020831.4		4-15	Susceptibility to severe bacterial infection Mild thrombocytopenia Congenital defects of phagocyte number or function
MMAA	18871	NM_172250.2		2-7	Methylmalonic aciduria, vitamin B12-responsive OMIM
MMAB	19331	NM_052845.3		1-9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type OMIM
MMADHC	25221	NM_015702.2		2-8	Methylmalonic aciduria and homocystinuria, cblD type OMIM
MOGS	24862	NM_006302.3		1-4	Congenital disorder of glycosylation, type IIb 606056 Bacterial and viral infections, severe neurologic disease, also known as congenital disorder of glycosylation type IIb (CDG-IIb) Predominantly Antibody Deficiencies
MPL	7217	NM_005373.2		1-12	Inherited Bone Marrow Failure Syndromes - Thrombocytopenia Congenital amegkaryocytic thrombocytopenia Congenital Amegakaryocytic Thrombocytopenia Amegakaryocytic Thrombocytopenia, Congenital Thrombocytopenia, congenital amegakaryocytic, 604498 Congenital amegakaryocytic thrombocytopenia (CAMT)
MPO	7218	NM_000250.1		1-12	Myeloperoxidase deficiency OMIM

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MRE11A	7230	NM_005591.3		2-20	Ataxia-telangiectasia-like disorder OMIM
MS4A1	7315	NM_152866.2		3-8	Immunodeficiency, common variable, 5 OMIM
MSH6	7329	NM_000179.2		1-10	Colorectal cancer, hereditary nonpolyposis, type 5 614350 Endometrial cancer, familial 608089 Mismatch repair cancer syndrome 276300 Family or personal history of cancer Predominantly Antibody Deficiencies
MSN	7373	NM_002444.2	11-13	1-13	Combined immunodeficiency Immunodeficiency 50, 300988 Recurrent infections with bacteria, varicella, neutropenia Immunodeficiencies affecting cellular and humoral immunity
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
MTR	7468	NM_000254.2		1-33	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	7473	NM_002454.2		2-15	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MUT	7526	NM_000255.3		2-13	Methylmalonic aciduria, mut(0) type OMIM
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
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
MYO5B	7603	NM_001080467.2	40	1-40	Microvillus inclusion disease 251850

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
MYSM1	29401	NM_001085487.2		1-20	MYSM1 deficiency Bone marrow failure neurodevelopmental delay mid-face hypoplasia immunodeficiency Short stature, recurrent infections, congenital bone marrow failure, myelodysplasia, immunodeficiency affecting B-cells and granulocytes, skeletal anomalies, cataracts, developmental delay. Combined immunodeficiencies with associated or syndromic features
NBAS	15625	NM_015909.3		1-52	Fever induced liver failure Infantile liver failure syndrome 2, 616483 Defects in Intrinsic and Innate Immunity
NBEAL2	31928	NM_015175.2		1-54	Gray platelet syndrome
NBN	7652	NM_002485.4		1-16	Nijmegen breakage syndrome OMIM
NCF1	7660	NM_000265.5	1-11	1-11	Chronic granulomatous disease due to deficiency of NCF-1 233700 Chronic granulomatous disease (CGD) Infections, autoinflammatory phenotype Congenital defects of phagocyte number or function
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
NCSTN	17091	NM_015331.2		1-17	Hidradenitis suppurativa with acne, 142690 Defects in Intrinsic and Innate Immunity familial hydradenitis suppurativa
NDNL2	7677	NM_138704.3		1	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 Severe lung disease (possibly viral), thymic hypoplasia, Chromosomal breakage, radiation sensitivity Combined immunodeficiencies with associated or syndromic features
NF1	7765	NM_000267.3	9-11, 13-29, 31-35	1-57	Myelodysplastic syndrome (MDS), Paediatric 162200
NFAT5	7774	NM_138714.3		5-15	NFAT5 haploinsufficieny IBD, recurrent sinopulmonary infections Diseases of Immune Dysregulation
NFKB1	7794	NM_003998.3		2-24	Immunodeficiency, common variable, 12 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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_021209.4		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
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
NOTCH1	7881	NM_017617.5		1-34	Acute lymphoblastic leukemia (ALL)
NPM1	7910	NM_002520.6		1-11	Acute myeloid leukaemia (AML)
NRAS	7989	NM_002524.4		2-5	Noonan syndrome 6 OMIM
NT5C3A	17820	NM_016489.12		2-10	Anemia, hemolytic, due to UMPH1 deficiency 266120
NUDT1	8048	NM_002452.3		2-4	Thrombocytopenia Absent-Radius Syndrome
OBFC1	26200	NM_024928.4		2-10	Intrauterine growth retardation, premature aging, pancytopenia, hypocellular bone marrow, gastrointestinal hemorrhage due to vascular ectasia, intracranial calcification, abnormal telomeres Combined immunodeficiencies with associated or syndromic features
OFD1	2567	NM_003611.2		1-23	Joubert syndrome 10 OMIM Simpson-Golabi-Behmel syndrome, type 2 OMIM
ORAI1	25896	NM_032790.3		1-2	Immunodeficiency 9 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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
PC	8636	NM_000920.3		3-22	Pyruvate carboxylase deficiency (ikke funnet relevant fenotype) OMIM
PDGFRA	8803	NM_006206.5		2-23	Hypereosinophilic syndrome, idiopathic, resistant to imatinib 607685
PDHA1	8806	NM_000284.3		1-11	Pyruvate dehydrogenase E1-alpha deficiency (ikke funnet relevant fenotype) OMIM
PDHX	21350	NM_003477.2		1-11	Lacticacidemia due to PDX1 deficiency (ikke funnet relevant fenotype) OMIM
PEPD	8840	NM_000285.3		1-15	Prolidase deficiency, 170100 Autoantibodies common, chronic skin ulcers, eczema, infections Diseases of Immune Dysregulation
PEX11B	8853	NM_003846.2		1-4	Thrombocytopenia Absent-Radius Syndrome
PFKM	8877	NM_000289.5		2-23	Glycogen storage disease VII, 232800
PGK1	8896	NM_000291.3		1-11	Phosphoglycerate kinase 1 deficiency OMIM
PGM3	8907	NM_001199917.1		2-14	Immunodeficiency 23 OMIM
PHF6	18145	NM_032458.2		2-10	T-cell acute lymphoblastic leukemia
PIAS3	16861	NM_006099.3		1-14	Thrombocytopenia Absent-Radius Syndrome
PIEZO1	28993	NM_001142864.4		1-51	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema OMIM
PIGA	8957	NM_002641.3	4-6	2-6	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Paroxysmal nocturnal haemoglobinuria, somatic, 300818
PIGT	14938	NM_015937.5		1-12	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIK3CD	8977	NM_005026.4	24	3-24	Immunodeficiency 14 OMIM
PIK3R1	8979	NM_181523.2		2-16	Immunodeficiency 36 OMIM ?Agammaglobulinemia 7, autosomal recessive OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
PKLR	9020	NM_000298.6		1-11	PYRUVATE KINASE DEFICIENCY Enzyme Disorder Pyruvate kinase deficiency, 266200
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
PMS2	9122	NM_000535.6	1-5, 9, 11-15	1-15	Mismatch repair cancer syndrome 276300 Post-Meiotic Segregation 2 (PMS2) deficiency CSR defects and Hyper IgM (HIGM) syndromes Recurrent infections, caf-au-lait spots, lymphoma, colorectal carcinoma, brain tumors Combined immunodeficiencies with associated or syndromic features
PNP	7892	NM_000270.3		1-6	Immunodeficiency due to purine nucleoside phosphorylase deficiency OMIM
POLA1	9173	NM_016937.3		1-37	Hyperpigmentation, characteristic facies, lung and GI involvement Autoinflammatory Disorders Pigmentary disorder, reticulate, with systemic manifestations, X-linked 301220 X-linked reticulate pigmentary disorder x-linked cutaneous amyloidosis with systemic features
POLE	9177	NM_006231.3		1-49	FILS syndrome OMIM
POLE2	9178	NM_002692.3		1-19	Recurrent infections, disseminated BCG infections, autoimmunity (type 1 diabetes, hypothyroidism, facial dysmorphism Combined immunodeficiencies with associated or syndromic features
POLR3GL	28466	NM_032305.2		2-8	Thrombocytopenia Absent-Radius Syndrome
PRF1	9360	NM_001083116.2		2-3	Aplastic anemia OMIM
PRKACG	9382	NM_002732.3		1	?Bleeding disorder, platelet-type, 19 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
PRKG1	9414	NM_006258.3		1-18	Enzyme Disorder

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
PROC	9451	NM_000312.3		2-9	Thrombophilia due to protein C deficiency, autosomal dominant OMIM Thrombophilia due to protein C deficiency, autosomal recessive OMIM
PSEN1	9508	NM_000021.3		3-12	Hidradenitis suppurative with cutaneous hyperpigmentation Defects in Intrinsic and Innate Immunity Acne inversa, familial, 3 613737
PSENEN	30100	NM_001281532.2		2-4	Hidradenitis suppurativa Defects in Intrinsic and Innate Immunity Acne inversa, familial, 2, with or without Dowling-Degos disease 613736
PSMA3	9532	NM_002788.3		1-11	CANDLE syndrome (Autoinflammation, lipodystrophy, and dermatosis syndrome)
PSMB4	9541	NM_002796.2		1-7	CANDLE syndrome (Autoinflammation, lipodystrophy, and dermatosis syndrome)
PSMB8	9545	NM_148919.3		1-6	Autoinflammation, lipodystrophy, and dermatosis syndrome OMIM
PSMB9	9546	NM_002800.4		1-6	Autoinflammation, lipodystrophy, and dermatosis syndrome CANDLE syndrome (Autoinflammation, lipodystrophy, and dermatosis syndrome)
PSTPIP1	9580	NM_003978.4		1-15	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne OMIM
PTEN	9588	NM_000314.6	9	1-9	Myelodysplastic syndrome (MDS), Adult Myelodysplastic syndrome (MDS), Paediatric Acute myeloid leukaemia (AML) Lymphoproliferation, Autoimmunity Predominantly Antibody Deficiencies
PTPN11	9644	NM_002834.4		1-15	LEOPARD syndrome 1 151100 Metachondromatosis 156250 Noonan syndrome 1 163950 Myelodysplastic syndrome (MDS), Paediatric Bleeding disorder LEOPARD syndrome, Leukemia, Juvenile myelomonocytic, somatic Metachondromatosis Noonan syndrome
PTPRC	9666	NM_002838.4		2-33	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive OMIM
PUS1	15508	NM_025215.5		1-6	Myopathy, Lactic Acidosis, and Sideroblastic Anemia, 600462
RAB27A	9766	NM_004580.4		2-6	Griscelli syndrome, type 2 OMIM
RAC2	9802	NM_002872.4		1-6	Neutrophil immunodeficiency syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
RAD21	9811	NM_006265.2	14	2-14	Acute myeloid leukaemia (AML) Especially in Down syndrome AML
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
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
RANBP2	9848	NM_006267.4	1-21	1-29	Fever induces acute encephalopathy Defects in Intrinsic and Innate Immunity
RASGRP1	9878	NM_005739.3		1-17	Recurrent pneumonia, herpesvirus infections, EBV associated lymphoma Diseases of Immune Dysregulation EBV-induced lymphoma Immunodeficiency Immunodeficiency 64, 618534 Immunodeficiency immunde dysregulation EBV-induced lymphoma
RBCK1	15864	NM_031229.3		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
RELB	9956	NM_006509.3		1-11	?Immunodeficiency 53, 617585 Recurrent infections Immunodeficiencies affecting cellular and humoral immunity
REN	9958	NM_000537.3		1-10	Hyperuricemic nephropathy, familial juvenile 2 OMIM
RET	9967	NM_020975.5		1-20	Central hypoventilation syndrome, congenital 209880

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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
RHCE	10008	NM_020485.5	1-10	1-10	Rh-null disease, amorph type OMIM
RHOH	686	NM_004310.4		3	Kronisk epidermodysplasia verruciformis/ oeket disposisjon for HPV PubMed
RIPK1	10019	NM_003804.5		2-11	Immunodeficiency 57, 618108 Severe immunodeficiency, arthritis, and intestinal inflammation
RLTPR	27089	NM_001013838.2		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
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_000975.5		1-6	Diamond-Blackfan anemia 7 OMIM
RPL13	10303	NM_001243130.1		2-6	Diamond-Blackfan anaemia
RPL15	10306	NM_002948.4	2-4	2-4	?Diamond-Blackfan anemia 12 OMIM
RPL18	10310	NM_000979.3		1-7	Diamond-Blackfan anaemia
RPL19	10312	NM_000981.4		1-6	Diamond-Blackfan anemia

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
RPL26	10327	NM_000987.5		2-4	?Diamond-Blackfan anemia 11 OMIM
RPL27	10328	NM_000988.5		2-5	Diamond-Blackfan anemia ?Diamond-Blackfan anemia 16, 617408
RPL31	10334	NM_001098577.2		2-5	Cytopenia - Diamond-Blackfan anaemia
RPL35	10344	NM_007209.3		1-4	Diamond-Blackfan anaemia
RPL35A	10345	NM_000996.4		2-5	Diamond-Blackfan anemia 5 OMIM
RPL5	10360	NM_000969.5		1-8	Diamond-Blackfan anemia 6 OMIM
RPL9	10369	NM_000661.4		2-7	Diamond-Blackfan anemia
RPS10	10383	NM_001014.5		2-6	Diamond-Blackfan anemia 9 OMIM
RPS14	10387	NM_001025070.1		2-5	Macrocyticanemia,refractory,dueto5qdeletion,somatic,153550 3 Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 -3
RPS17	10397	NM_001021.6	1-5	1-5	Diamond-Blackfan anemia 4 612527
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_001029.5		1-4	Diamond-Blackfan anemia 10 OMIM
RPS27	10416	NM_001030.6		1-4	Diamond-Blackfan anemia ?Diamond-Blackfan anemia 17, 617409
RPS28	10418	NM_001031.4		1-3	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	10419	NM_001032.4		1-3	Diamond-Blackfan anemia 13 OMIM
RPS7	10440	NM_001011.4		2-7	Diamond-Blackfan anemia 8 OMIM
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

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RUNX1	10471	NM_001754.4		2-9	Platelet disorder, familial, with associated myeloid malignancy OMIM
SALL4	15924	NM_020436.4		1-4	IVIC syndrome OMIM
SAMD9	1348	NM_017654.3		3	MIRAGE syndrome, 617053 MIRAGE syndrome 617053 IUGR with gonadal abnormalities, adrenal failure, MDS with chromosome 7 aberrations, predisposition to infections, enteropathy, absent spleen Combined immunodeficiencies with associated or syndromic features MIRAGE syndrome (Myelodysplasia, Infection, Restriction of growth, Adrenal insufficiency, Genital phenotypes, and Enteropathy) ataxia-thrombocytopenia syndrome
SAMD9L	1349	NM_152703.4		5	Ataxia-pancytopenia syndrome 159550 Cytopenia, predisposition to MDS with chromosome 7 aberrations, immunodeficiency, and progressive cerebellar dysfunction Combined immunodeficiencies with associated or syndromic features
SAMHD1	15925	NM_015474.3		1-16	Aicardi-Goutieres syndrome 5 OMIM
SART3	16860	NM_014706.3		1-19	Porokeratosis
SBDS	19440	NM_016038.3	1-5	1-5	Shwachman-Diamond syndrome OMIM
SEC23B	10702	NM_006363.6		2-20	Congenital dyserythropoietic anemia type II Congenital Dyserythropoietic Anemia Anemia, dyserythropoieticcongenital, type II, 224100 ANEMIA, DYSERYTHROPOIETIC CONGENITAL, TYPE II
SEMA3E	10727	NM_012431.2		1-17	Charge syndrome 214800 CHARGE syndrome immune-mediated cerebellar ataxia Coloboma, heart anomaly, choanal atresia, intellectual retardation, genital and ear anomalies, CNS malformation, some are SCID-like and have low TRECs Combined immunodeficiencies with associated or syndromic features
SERPING1	1228	NM_000062.2		2-8	Complement component 4, partial deficiency of OMIM
SETBP1	15573	NM_015559.3		2-6	Myelodysplastic syndrome (MDS), Paediatric
SF3B1	10768	NM_012433.3		1-25	Myelodysplastic syndrome, somatic, 614286

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SGPL1	10817	NM_003901.3		2-15	Nephrotic syndrome 14, 617575 drenal insufficiency steroid-resistant nephrotic syndrome focal segmental glomerulosclerosis lymphopenia
SH2D1A	10820	NM_002351.4		1-4	Lymphoproliferative syndrome, X-linked, 1 OMIM
SH3BP1	10824	NM_018957.5		1-18	Myelodysplastic syndrome (MDS), Paediatric
SH3BP2	10825	NM_003023.4		2-13	Cherubism 118400 Other autoinflammatory diseases with known genetic defect Bone degeneration in jaws Autoinflammatory Disorders
SKIV2L	10898	NM_006929.4		1-28	Trichohepatoenteric syndrome 2 OMIM
SLC11A2	10908	NM_000617.2		2-16	Anemia, hypochromic microcytic, with iron overload 1 206100
SLC19A2	10938	NM_006996.2		1-6	Thiamine-Responsive Megaloblastic Anemia syndrome 249270
SLC25A38	26054	NM_017875.4		1-7	Anemia, sideroblastic, 2, pyridoxine-refractory 205950
SLC29A3	23096	NM_018344.5		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM
SLC2A1	11005	NM_006516.3		1-10	Stomatocytosis Pyridoxine-refractory sideroblastic anemia
SLC34A1	11019	NM_003052.4		2-13	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Fanconi renotubular syndrome 2, 613388
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
SLC4A1	11027	NM_000342.3		2-20	Haemolytic Anemia RBC membrane abnormality Cryohydrocytosis,185020 Ovalocytosis, SA type, 166900 Spherocytosis, type 4, 612653
SLC7A7	11065	NM_001126106.2		3-11	Lysinuric protein intolerance OMIM
SLFN14	32689	NM_001129820.1		1-4	SLFN14-related thrombocytopenia Platelet disorder Bleeding Disorder, platelet-type, 20

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SLX4	23845	NM_032444.3		2-15	Fanconi anemia, complementation group P OMIM
SMARCAL1	11102	NM_014140.3		3-18	Schimke immunoosseous dysplasia OMIM
SMARCD2	11107	NM_001098426.1		1-13	Neutropenia, developmental aberrations, skeletal abnormalities, hematopoietic stem cells, myelodysplasia Congenital defects of phagocyte number or function
SMC1A	11111	NM_006306.3		1-25	Acute Promyelocytic Leukemia Acute myeloid leukaemia (AML) Especially in Down syndrome AML
SMC3	2468	NM_005445.3		1-29	Acute myeloid leukaemia (AML) Especially in Down syndrome AML
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
SPINK5	15464	NM_006846.3		1-33	Netherton syndrome OMIM Atopy OMIM
SPPL2A	30227	NM_032802.3		1-15	Defects with susceptibility to mycobacterial infection (MSMD) Susceptibility to mycobacteria Defects in Intrinsic and Innate Immunity
SPTA1	11272	NM_003126.3		1-52	RBC membrane abnormality Elliptocytosis-2 (MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown), 130600 Pyropoikilocytosis (BIALLELIC, autosomal or pseudoautosomal), 266140 Spherocytosis, type 3 (BIALLELIC, autosomal or pseudoautosomal), 270970
SPTB	11274	NM_001024858.3		1-35	RBC membrane abnormality Elliptocytosis Spherocytosis,616649 Anemia, neonatal hemolytic, fatal and near-fatal
SRP54	11301	NM_003136.3		2-16	Schwachman Diamond features Congenital defects of phagocyte number or function
SRP72	11303	NM_006947.4	19	1-19	Bone marrow failure syndrome 1 OMIM
SRSF2	10783	NM_003016.4		1-2	Myelodysplastic syndrome (MDS), Paediatric
STAG2	11355	NM_001042749.2		3-35	Acute myeloid leukaemia (AML) Especially in Down syndrome AML

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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
STAT5A	11366	NM_003152.3	7-10	3-20	Combined immunodeficiency Defects with susceptibility to mycobacterial infection (MSMD)
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_006282.4		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
TAZ	11577	NM_000116.4		1-11	Barth syndrome OMIM
TBK1	11584	NM_013254.3		2-21	Herpes simplex encephalitis, susceptibility to Herpetic encephalitis (HSE) {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8} 617900 Herpes simplex virus 1 encephalitis Defects in Intrinsic and Innate Immunity
TBX1	11592	NM_080647.1		2-9	DiGeorge syndrome OMIM Velocardiofacial syndrome OMIM
TBXAS1	11609	NM_001061.4		1-13	Ghosal syndrome
TCF3	11633	NM_003200.4		2-19	Agammaglobulinemia 8, autosomal dominant OMIM

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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
TET2	25941	NM_001127208.2		3-11	Myelodysplastic syndrome, somatic, 614286
TF	11740	NM_001063.3		1-17	Atransferrinemia, 209300 Congenital hypotransferrinemia
TFR2	11762	NM_003227.3		1-18	Hemochromatosis, type 3 OMIM
TFRC	11763	NM_003234.3		2-19	Immunodeficiency 46 OMIM
THBD	11784	NM_000361.2		1	{Hemolytic uremic syndrome, atypical, susceptibility to, 6} OMIM
THPO	11795	NM_000460.4		2-6	Thrombocytopenia and thrombocythemia 1
TICAM1	18348	NM_182919.3		2	{Herpes simplex encephalitic, susceptibility to, 6} OMIM
TINF2	11824	NM_001099274.2		1-9	Dyskeratosis congenita, autosomal dominant 3 OMIM Revesz syndrome OMIM
TIRAP	17192	NM_148910.2		4-5	Defects of TLR/NFkappa-B signalling TIRAP deficiency Staphylococcal disease during childhood Defects in Intrinsic and Innate Immunity
TLR3	11849	NM_003265.2		2-5	{Herpes simplex encephalitis, susceptibility to, 2} OMIM
TMC6	18021	NM_007267.7		2-20	Epidermodysplasia verruciformis OMIM
TMC8	20474	NM_152468.4		2-16	Epidermodysplasia verruciformis OMIM
TMEM173	27962	NM_198282.3		3-8	STING-associated vasculopathy, infantile-onset OMIM
TMPRSS6	16517	NM_153609.3		1-18	Iron-Refractory Iron Deficiency Anemia Iron refractoryirondeficiencyanemia,206200

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TNFAIP3	11896	NM_006290.3		2-9	A20 deficiency Autoinflammatory syndrome, familial, Behcet-like, 616744 Autoimmune lymphoproliferative syndrome Arthralgia, mucosal ulcers, ocular inflammation Autoinflammatory Disorders
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
TPI1	12009	NM_000365.5		1-7	Hemolytic anemia due to triosephosphate isomerase deficiency,615512 Enzyme Disorder
TPP2	12016	NM_003291.3		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_033629.5		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
TSR2	25455	NM_058163.2		1-5	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTC37	23639	NM_014639.3		4-43	Trichohepatoenteric syndrome 1 OMIM
TTC7A	19750	NM_020458.3		1-20	Gastrointestinal defects and immunodeficiency syndrome OMIM
TUBB1	16257	NM_030773.3		1-4	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112 Macrothrombocytopenia, Beta-tubulin 1 related

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TXNIP	16952	NM_006472.5		1-8	Thrombocytopenia Absent-Radius Syndrome
TYK2	12440	NM_003331.4		3-25	Immunodeficiency 35 OMIM
U2AF1	12453	NM_006758.2		1-8	Acute myeloid leukaemia (AML) Myelodysplastic syndrome (MDS)
UBE2T	25009	NM_014176.4		2-7	Falcon anemia Fanconi anemia, complementation group T, 616435
UNC119	12565	NM_005148.3		1-5	?Immunodeficiency 13 OMIM
UNC13D	23147	NM_199242.2		1-32	Hemophagocytic lymphohistiocytosis, familial, 3 OMIM
UNC93B1	13481	NM_030930.3	10-12	1-12	Herpes simplex encephalitis, susceptibility to, 1 Herpetic encephalitis (HSE) {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1} 610551 Herpes simplex virus 1 encephalitis Defects in Intrinsic and Innate Immunity
UNG	12572	NM_080911.2		1-7	Immunodeficiency with hyper IgM, type 5 OMIM
UROS	12592	NM_000375.2		2-10	Porphyria, congenital erythropoietic 263700
USB1	25792	NM_024598.3		1-7	Poikiloderma med neutropeni OMIM
USP18	12616	NM_017414.3	3-11	2-11	Pseudo-TORCH syndrome 2, 617397 (includes Thrombocytopenia) TORCH like syndrome Autoinflammatory Disorders Pseudo-TORCH syndrome 2, 617397
VPS13B	2183	NM_017890.4		2-62	Cohen syndrome OMIM
VPS45	14579	NM_007259.5		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
WDR1	12754	NM_017491.4		1-15	Mild neutropenia, poor wound healing, severe stomatitis, neutrophil nuclei herniate Congenital defects of phagocyte number or function
WIPF1	12736	NM_001077269.1		2-8	?Wiskott-Aldrich syndrome 2 OMIM

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WRAP53	25522	NM_018081.2		1-10	Dyskeratosis congenita, autosomal recessive 3 OMIM
WT1	12796	NM_024426.5		1-10	Acute myeloid leukaemia (AML)
XIAP	592	NM_001167.3	7	2-7	Lymphoproliferative syndrome, X-linked, 2 OMIM
XRCC2	12829	NM_005431.1		1-3	?Fanconi anemia, complementation group U OMIM
YARS2	24249	NM_001040436.2		1-5	Myopathy, lactic acidosis, and sideroblastic anemia 2 613561
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
ZNF341	15992	NM_032819.4	15	1-15	Hyper-IgE recurrent infection syndrome 3, autosomal recessive OMIM
ZRSR2	23019	NM_005089.3		1-11	Acute myeloid leukaemia (AML) Chronic Myeloid Leukemia (CML)