Oslo universitetssykehus
Avdeling for medisinsk genetikk

Tidlig debuterende inflammatorisk tarmsykdom

Genpanel, versjon v01

* 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: **NCF1**

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
ADA	186	NM_000022.4		1-12	Severe combined immunodeficiency due to ADA deficiency (SCID) 102700 OMIM
ADAM17	195	NM_003183.6		1-19	ADAM-17 deficiency OMIM Inflammatory skin and bowel disease, neonatal, 1 614328 OMIM
AICDA	13203	NM_020661.4		1-5	Immunodeficiency with hyper-IgM, type 2 605258 OMIM
BTK	1133	NM_000061.2		2-19	Agammaglobulinemia and isolated hormone deficiency 307200 OMIM Agammaglobulinemia, X-linked 1 300755 OMIM
CD3G	1675	NM_000073.2		1-6	Immunodeficiency 17, CD3 gamma deficient 615607 OMIM

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CD40LG	11935	NM_000074.3		1-5	Immunodeficiency, X-linked, with hyper-IgM 308230 OMIM
COL7A1	2214	NM_000094.3		1-118	Dystrophic epidermolysis bullosa OMIM
CTLA4	2505	NM_005214.5		1-4	CTLA4 deficiency OMIM
CYBA	2577	NM_000101.4		1-6	Chronic granulomatous disease, autosomal, due to deficiency of CYBA 233690 OMIM
CYBB	2578	NM_000397.4		1-13	Chronic granulomatous disease, X-linked 306400 OMIM
DCLRE1C	17642	NM_001033855.3	4-9	1-14	Omenn syndrome 603554 OMIM Severe combined immunodeficiency, Athabascan type 602450 OMIM
DKC1	2890	NM_001363.5		1-15	Hoyeraal Hreidarsson Syndrome OMIM Dyskeratosis congenita, X-linked OMIM
DOCK8	19191	NM_203447.3		1-48	Hyper-IgE recurrent infection syndrome, autosomal recessive 243700 OMIM
EPCAM	11529	NM_002354.3		1-9	Diarrhea 5, with tufting enteropathy, congenital 613217 OMIM
FAM105B	25118	NM_138348.6		1-7	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 OMIM AIPDS OMIM
FERMT1	15889	NM_017671.4		2-15	Kindler syndrome OMIM
FOXP3	6106	NM_014009.4		2-12	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked (IPEX) 304790 OMIM
G6PC3	24861	NM_138387.3		1-6	Congenital neutropenia OMIM
GUCY2C	4688	NM_004963.4		1-27	Familial Diarrhea 6 614616 OMIM Meconium ileus OMIM
HPS1	5163	NM_000195.5	4-6	3-20	Hermansky-Pudlak syndrome 1 203300 OMIM

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HPS4	15844	NM_022081.5		2-14	Hermansky-Pudlak syndrome 4 614073 OMIM
HPS6	18817	NM_024747.6		1	Hermansky-Pudlak syndrome 6 614075 OMIM
ICOS	5351	NM_012092.4		1-5	Immunodeficiency, common variable, 1 607594 OMIM
IKBKG	5961	NM_001099857.3	3-10	2-10	Ectodermal X-linked dysplasia, hypohidrotic, with immune deficiency 300291 OMIM
IL10RA	5964	NM_001558.3		1-7	IL-10 signalling defects / deficiency OMIM Inflammatory bowel disease 28, early onset, autosomal recessive 613148 OMIM
IL10RB	5965	NM_000628.5		1-7	IL-10 signalling defects / deficiency OMIM Inflammatory bowel disease 25, early onset, autosomal recessive 612567 OMIM
IL21	6005	NM_021803.4		1-5	IL21 deficiency (Combined variable immunodeficiency-like) OMIM severe diarrhea and inflammatory bowel disease OMIM Early-onset inflammatory bowel disease OMIM
IL2RA	6008	NM_000417.3		1-8	Immunodeficiency 41 with lymphoproliferation and autoimmunity (IPEX-like) 606367 OMIM
IL2RG	6010	NM_000206.2		1-8	Severe combined immunodeficiency, X-linked 300400 OMIM
ITGB2	6155	NM_000211.5		2-16	Leukocyte adhesion deficiency 116920 OMIM
LIG4	6601	NM_002312.3		2	SCID OMIM LIG4 syndrome 606593 OMIM
LRBA	1742	NM_006726.4		2-58	Immunodeficiency, common variable, 8, with autoimmunity (CVID 8) 614700 OMIM
MEFV	6998	NM_000243.2		1-10	Familial Mediterranean fever, AD 134610 OMIM Familial Mediterranean fever, AR 249100 OMIM

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MVK	7530	NM_000431.4		2-11	Hyper-IgD syndrome 260920 OMIM Mevalonic aciduria 610377 OMIM
NCF1	7660	NM_000265.6	1-11	1-11	Chronic granulomatous disease due to deficiency of NCF-1 233700 OMIM
NCF2	7661	NM_000433.3		1-15	Chronic granulomatous disease due to deficiency of NCF-2 233710 OMIM
NPC1	7897	NM_000271.5		1-25	Niemann_Pick type C disease OMIM Niemann-Pick disease, type D OMIM
PIK3CD	8977	NM_005026.5	24	3-24	PI3K activation syndrome OMIM Immunodeficiency 14 615513 OMIM
PIK3R1	8979	NM_181523.3		2-16	Agammaglobulinemia 7, autosomal recessive 615214 OMIM
PLCG2	9066	NM_002661.5		2-33	Phospholipase C_2 defects OMIM Autoinflammation, antibody deficiency, and immune dysregulation syndrome 614878 OMIM
PTEN	9588	NM_000314.8	9	1-9	PTEN hamartoma tumor syndrome OMIM Bannayan-Riley-Ruvalcaba syndrome 153480 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_000536.4		2	SCID/Hyper-IgM OMIM Severe combined immunodeficiency, B cell-negative 601457 OMIM Omenn syndrome 603554 OMIM

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RTEL1	15888	NM_032957.5		2-35	Hoyeraal Hreidarsson Syndrome OMIM Dyskeratosis congenita, autosomal dominant 4 OMIM Dyskeratosis congenita, autosomal recessive 5 OMIM
SAMD9	1348	NM_017654.4		3	MIRAGE syndrome 617053 OMIM
SH2D1A	10820	NM_002351.4		1-4	Lymphoproliferative syndrome, X-linked, 1 308240 OMIM
SKIV2L	10898	NM_006929.5		1-28	Trichohepatoenteric syndrome 2 614602 OMIM
SLC37A4	4061	NM_001164277.1		3-11	Glycogen storage disease type 1b 232220 OMIM
STAT1	11362	NM_007315.3		3-25	IPEX-like OMIM Immunodeficiency 31A, mycobacteriosis, autosomal dominant 614892 OMIM Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive 613796 OMIM Immunodeficiency 31C, autosomal dominant 614162 OMIM
STAT3	11364	NM_139276.2		2-24	Autoimmune disease, multisystem, infantile-onset, 1 OMIM
STXBP2	11445	NM_006949.4		1-19	Hemophagocytic lymphohistiocytosis, familial, 5 613101 OMIM
TGFBR1	11772	NM_004612.4		1-9	Loeys-Dietz Syndrome OMIM
TGFBR2	11773	NM_003242.6		1-7	Loeys-Dietz Syndrome 2 OMIM
TTC37	23639	NM_014639.3		4-43	Trichohepatoenteric syndrome 1 222470 OMIM

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TTC7A	19750	NM_020458.4		1-20	TTC7A deficiency OMIM Epithelial Barrier Dysfunction OMIM Gastrointestinal defects and immunodeficiency syndrome 243150 OMIM
WAS	12731	NM_000377.3		1-12	Wiskott-Aldrich syndrome 301000 OMIM
XIAP	592	NM_001167.3	7	2-7	Lymphoproliferative syndrome, X-linked, 2 300635 OMIM
ZAP70	12858	NM_001079.3		3-14	SCID OMIM Autoimmune disease, multisystem, infantile-onset, 2 617006 OMIM