Oslo universitetssykehus
Avdeling for medisinsk genetikk

Febersykdommer

Genpanel, versjon v02

* 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).

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
CARD14	16446	NM_024110.4		2-21	Pityriasis rubra pilaris OMIM Psoriasis 2 OMIM
CDC42	1736	NM_001791.3	4-6	2-6	Takenouchi-Kosaki syndrome OMIM
CECR1	1839	NM_001282228.1		2-10	?Sneddon syndrome OMIM Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome OMIM
ELANE	3309	NM_001972.2		1-5	Neutropenia, cyclic OMIM Neutropenia, severe congenital 1, autosomal dominant OMIM
F12	3530	NM_000505.3		1-14	Angioedema, hereditary, type III OMIM
FAM105B	25118	NM_138348.4		1-7	Autoinflammation, panniculitis, and dermatosis syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
IKBKG	5961	NM_001099856.3	3-10	1-10	Ectodermal dysplasia and immunodeficiency 1 OMIM Immunodeficiency 33 OMIM Incontinentia pigmenti OMIM
IL1RN	6000	NM_173843.2		3-6	Interleukin 1 receptor antagonist deficiency OMIM
IL36RN	15561	NM_173170.1		2-5	Psoriasis 14, pustular OMIM
LACC1	26789	NM_001128303.1		2-6	Juvenile arthritis OMIM
LPIN2	14450	NM_014646.2		2-20	Majeed syndrome OMIM
MEFV	6998	NM_000243.2		1-10	Familial Mediterranean fever, AD OMIM Familial Mediterranean fever, AR OMIM Neutrophilic dermatosis, acute febrile OMIM
MVK	7530	NM_001114185.2		2-11	Hyper-IgD syndrome OMIM Mevalonic aciduria OMIM Porokeratosis 3, multiple types OMIM
NCSTN	17091	NM_015331.2		1-17	Acne inversa, familial, 1 OMIM
NLRC4	16412	NM_001199139.1		2-9	?Familial cold autoinflammatory syndrome 4 OMIM Autoinflammation with infantile enterocolitis OMIM
NLRP1	14374	NM_001033053.3		1-16	?Respiratory papillomatosis, juvenile recurrent, congenital OMIM Autoinflammation with arthritis and dyskeratosis OMIM Palmoplantar carcinoma, multiple self-healing OMIM
NLRP12	22938	NM_144687.3		1-10	Familial cold autoinflammatory syndrome 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
NLRP3	16400	NM_001079821.2		3-11	CINCA syndrome OMIM Deafness, autosomal dominant 34, with or without inflammation OMIM Familial cold inflammatory syndrome 1 OMIM Keratoendothelitis fugax hereditaria OMIM Muckle-Wells syndrome OMIM
NOD2	5331	NM_022162.2		1-12	Blau syndrome OMIM
PLCG2	9066	NM_002661.4		2-33	Autoinflammation, antibody deficiency, and immune dysregulation syndrome OMIM Familial cold autoinflammatory syndrome 3 OMIM
POMP	20330	NM_015932.6		1-6	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma OMIM Proteasome-associated autoinflammatory syndrome 2 OMIM PubMed
PSMA3	9532	NM_152132.2		1-11	proteasome-associated autoinflammatory syndrome PubMed
PSMB10	9538	NM_002801.3		1-8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms PubMed
PSMB4	9541	NM_002796.2		1-7	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms OMIM
PSMB8	9545	NM_004159.5		1-6	Proteasome-associated autoinflammatory syndrome 1 and digenic forms OMIM
PSMB9	9546	NM_002800.4		1-6	?Proteasome-associated autoinflammatory syndrome 3, digenic OMIM
PSMG2	24929	NM_147163.1		2-7	Proteasome-associated autoinflammatory syndrome 4 PubMed

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
PSTPIP1	9580	NM_003978.3		1-15	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne OMIM
RBCK1	15864	NM_031229.2		1-12	Polyglucosan body myopathy 1 with or without immunodeficiency OMIM
RIPK1	10019	NM_003804.3		1-10	Autoinflammation with episodic fever and lymphadenopathy OMIM Immunodeficiency 57 with autoinflammation OMIM
SAMD9L	1349	NM_152703.5		5	Ataxia-pancytopenia syndrome OMIM Monosomy 7 myelodysplasia and leukemia syndrome 1 OMIM
SLC29A3	23096	NM_018344.6		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM
TMEM173	27962	NM_198282.3		3-8	STING-associated vasculopathy, infantile-onset OMIM
TNFAIP3	11896	NM_001270507.2		2-9	Autoinflammatory syndrome, familial, Behcet-like OMIM
TNFRSF11A	11908	NM_001278268.2		1-10	{Paget disease of bone 2, early-onset} OMIM Osteolysis, familial expansile OMIM Osteopetrosis, autosomal recessive 7 OMIM
TNFRSF1A	11916	NM_001065.3		1-10	Periodic fever, familial OMIM
TRNT1	17341	NM_182916.2		2-8	Retinitis pigmentosa and erythrocytic microcytosis OMIM Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay OMIM
WDR1	12754	NM_017491.3		1-15	Periodic fever, immunodeficiency, and thrombocytopenia syndrome OMIM