Locus 174 (rs6916921)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,520,426-31,563,240
block size	42,815 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.904 [-2.113 – 0.076]	gene(s) hit or close-by	ATP6V1G2, LST1, LTA, LTB, NCR3, NFKBIL1, TNF
phastCons	μ = 0.014 [0 – 0.08]	eQTL gene(s)	AIF1, ATP6V1G2, BAG6, HLA-B, LST1, MICA, MICB, NCR3, PSORS1C1, XXbac-BPG181B23.7, XXbac-BPG299F13.17
GERP++	μ = -1.943 [-7.11 – 0.109]	potentially regulated gene(s)	AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, C2, C2, C2, C2, C2, C2, C2, C2, C2, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CYP21A1P, CYP21A1P, CYP21A1P, CYP21A1P, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICA, MICA, MICA, MICA, MICA, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, STK19, STK19, STK19, STK19, STK19, STK19, STK19, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.12
CADD score	μ = 3.095 [0.301 – 4.713]	disease gene(s)	NFKBIL1, TNF, LTA, NCR3, C2, HLA-B

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR993387	1
?	HGMD curated	HGMD	CR082032	1
VASCULAR DEMENTIA, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:191160	1

Disease gene annotation

gene	trait	source DB	source entry/link
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507

	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ATP6V1C2	ENST00000481908	ILMN_1654541	skin	6.39×10-8 (p-value)	MuTHER consortium	3

Locus 174 (sentinel SNP rs6916921 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

ATP6V1G2	ENST00000481998	ILMN_1654541	skin	6.20×10-9 (p-value)	MuTHER consortium	3
ATP6V1G2	ENST00000303892		blood	1.36×10-6 (p-value)	MuTHER consortium	3
ATP6V1G2	ENST00000483170		adipocyte	2.03×10-5 (p-value)	MuTHER consortium	3
ATP6V1G2	ENST00000376151					
AIF1	ENST00000337917	ILMN_1792473	blood	1.13×10-8 (p-value)	MuTHER consortium	3
AIF1	ENST00000466820					
AIF1	ENST00000497362					
AIF1	ENST00000376059					
AIF1	ENST00000376049					
BAG6	?	ENSG00000204463	muscularis mucosae	2.06×10-9 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	lung	6.51×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	transformed fibroblasts	1.48×10-9 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	transformed fibroblasts	2.17×10-7 (p-value)	GTEx Portal V6	6
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	6.61×10-7 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	tibial artery	5.46×10-8 (p-value)	GTEx Portal V6	6
LST1	ENST00000303757	ILMN_1718936	monocyte	4.42×10-5 (p-value)	Fairfax et al.	4
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
MICB	ENST00000399150	ILMN_1708006	monocyte	8.70×10-5 (p-value)	Fairfax et al.	3
MICB	ENST00000538442		b-cell	6.58×10-4 (p-value)	Fairfax et al.	3
MICB	ENST00000252229					
MICA	?	ENSG00000204520	thyroid	4.01×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	skeletal muscle	2.55×10-8 (p-value)	GTEx Portal V6	6
MICB	?	ENSG00000204516	blood	3.91×10-3 (q-value)	SeeQTL DB (HapMap)	2
NCR3	?	ENSG00000204475	blood	9.04×10-5 (q-value)	SeeQTL DB (HapMap)	1
MICA	?	ENSG00000204520	sun exposed skin	2.35×10-10 (p-value)	GTEx Portal V6	6
PSORS1C1	?	ENSG00000204540	sun exposed skin	2.60×10-6 (p-value)	GTEx Portal V6	4
BAG6	?	ENSG00000204463	sun exposed skin	2.55×10-6 (p-value)	GTEx Portal V6	3
MICA	?	ENSG00000204520	unexposed skin	6.49×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	aorta	1.94×10-6 (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	subcutaneous adipocytes	9.87×10-8 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	subcutaneous adipocytes	7.18×10-9 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	visceral adipocytes	8.77×10-8 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	cortex	3.07×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	tibial nerve	1.35×10-8 (p-value)	GTEx Portal V6	6

MICA	?	ENSG00000204520	tibial nerve	1.35×10-8 (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	tibial nerve	1.10×10-5 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sigmoid colon	6.30×10-7 (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	esophagus mucosa	3.60×10-8 (p-value)	GTEx Portal V6	6
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	5.43×10-6 (p-value)	GTEx Portal V6	5
BAG6	ENST00000375976	ILMN_1705364	blood	4.62×10-6 (p-value)	Westra et al.	2
BAG6	ENST00000362049					
BAG6	ENST00000375964					
BAG6	ENST00000464869					
BAG6	ENST00000422948					
BAG6	ENST00000211379					
BAG6	ENST00000441793					
BAG6	ENST00000439687					
BAG6	ENST00000464126					
BAG6	?	ENSG00000204463	cerebellum	9.53×10-9 (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444842	1	ENCP00000048041	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048046	PRRC2A PRRC2A

	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048109	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048051	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048150	STK19 STK19 STK19 STK19 STK19 STK19
ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048033	TNF TNF TNF TNF TNF TNF TNF TNF
ENCP00000048035	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048116	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
ENCP00000048043	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048048	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A

	PRRC2A
ENCP00000048093	MSH5 MSH5 MSH5 MSH5 MSH5 MSH5 MSH5 MSH5
ENCP00000048006	XXbac-BPG248L24.12
ENCP00000048134	C2 C2 C2 C2 C2 C2 C2 C2
ENCP00000048037	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
ENCP00000048002	XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10
ENCP00000048038	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3

ENCP00000048005	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048017	HCP5 HCP5 HCP5 HCP5
ENCP00000048023	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
ENCP00000047991	HCG27 HCG27 HCG27 HCG27 HCG27 HCG27 HCG27
ENCP00000048104	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048052	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048110	VARS VARS VARS VARS VARS VARS VARS
ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048157	CYP21A1P CYP21A1P CYP21A1P CYP21A1P
ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1

	ENCP00000048040	UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487867 (CTCF binding site)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac
		blood (K562)	CTCF
		monocytes (Monocytes-CD14+)	H3K4me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	BATF, DNase1, CTCF, H3K79me2, H3K27ac, H3K4me2, H3K9ac, PolII, EBF1
		lung (IMR90)	H3K27me3
ENSR00001494573 (promoter)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K27me3, H3K36me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	EBF1, POU2F2, Cmyc, PolII, DNase1, Egr1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me2
ENSR00000264419 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K4me2, H3K4me3, H4K20me1, H3K36me3
		liver (HepG2)	H3K27me3
		blood (K562)	H2AZ

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATP6V1G2	upstream gene variant	4222	ENST00000415099	?	ENSP00000390148	1
LST1	downstream gene variant	24	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	256	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	122	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	175	ENST00000460834	?	?	1
LST1	downstream gene variant	122	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	23	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	1091	ENST00000419073	?	?	1
LST1	downstream gene variant	175	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	23	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	23	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	23	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	23	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	270	ENST00000376092	?	ENSP00000365260	1
LST1	downstream gene variant	23	ENST00000303757	NM_205840.2	ENSP00000303649	1

LST1	downstream gene variant	23	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	23	ENST00000464526	?	?	1
LST1	downstream gene variant	306	ENST00000396112	?	ENSP00000379418	1
LST1	downstream gene variant	23	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	1202	ENST00000433492	?	ENSP00000399869	1
LST1	downstream gene variant	23	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	175	ENST00000464044	?	ENSP00000417645	1
LTA	downstream gene variant	381	ENST00000489638	?	?	2
LTA	downstream gene variant	381	ENST00000454783	NM_001159740.2	ENSP00000403495	2
LTA	downstream gene variant	381	ENST00000418386	NM_000595.3	ENSP00000413450	2
LTA	downstream gene variant	381	ENST00000471842	?	?	2
LTB	downstream gene variant	4505	ENST00000429299	NM_002341.1	ENSP00000410481	1
LTB	downstream gene variant	4574	ENST00000482429	?	?	1
LTB	downstream gene variant	4505	ENST00000446745	NM_009588.1	ENSP00000416113	1
LTB	downstream gene variant	4475	ENST00000483972	?	?	1
NCR3	downstream gene variant, upstream gene variant	342	ENST00000376072	NM_001145467.1	ENSP00000365240	2
NCR3	downstream gene variant, upstream gene variant	365	ENST00000376071	?	ENSP00000365239	2
NCR3	upstream gene variant	2651	ENST00000495600	?	?	1
NCR3	upstream gene variant	2504	ENST00000491161	?	?	1
NCR3	upstream gene variant	2478	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	upstream gene variant	2478	ENST00000340027	NM_147130.2	ENSP00000342156	1
NFKBIL1	downstream gene variant	4415	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	downstream gene variant	4161	ENST00000473655	?	?	1
NFKBIL1	downstream gene variant	3291	ENST00000376148	NM_005007.3	ENSP00000365318	1
NFKBIL1	downstream gene variant	3291	ENST00000376145	NM_001144961.1	ENSP00000365315	1
NFKBIL1	downstream gene variant	3291	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1
TNF	upstream gene variant	862	ENST00000449264	NM_000594.3	ENSP00000398698	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFKBIL1	ENST00000473655	?	?	1
NFKBIL1	ENST00000376148	NM_005007.3	ENSP00000365318	1
NFKBIL1	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1
NFKBIL1	ENST00000376145	NM_001144961.1	ENSP00000365315	1
TNF	ENST00000449264	NM_000594.3	ENSP00000398698	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NCR3	ENST00000491161	?	1
NCR3	ENST00000495600	?	1