SNiPAcard

Block annotations

Block info

genomic range	chr11:73,090,265-73,293,082
block size	202,818 bp
variant count	32 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.377 [-4.432 – 4.671]	**gene(s) hit or close-by**	FAM168A, HMGN2P38, RELT, RP11-809N8.2, RP11-809N8.4, RP11-809N8.5, RP11-809N8.6
phastCons	μ = 0.110 [0 – 1]	**eQTL gene(s)**	ARHGEF17, FAM168A, RELT, RP11-809N8.4
GERP++	μ = 0.507 [-2.7 – 3.72]	**potentially regulated gene(s)**	ARHGEF17, P2RY2, P2RY6, RAB6A, RELT, RP11-800A3.2, RP11-809N8.4, RP11-809N8.5
CADD score	μ = 5.363 [0.576 – 21.4]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FAM168A	?	ENSG00000054965	blood	4.50×10^{-7} (p-value)	GTEx Portal V6	30
RP11-809N8.4	?	ENSG00000256448	blood	1.17×10^{-5} (p-value)	GTEx Portal V6	1
ARHGEF17	ENST00000543530	ILMN_1754562	monocyte	6.35×10^{-8} (p-value)	Fairfax et al.	3
ARHGEF17	ENST00000263674					
RELT	ENST00000064780	ILMN_1748614	monocyte	8.43×10^{-10} (p-value)	Fairfax et al.	3
RELT	ENST00000545886		b-cell	8.84×10^{-6} (p-value)	Fairfax et al.	3
RELT	ENST00000393580					
RELT	ENST00000539134					
ARHGEF17	ENST00000543530	ILMN_1754562	monocyte	3.23×10^{-15} (p-value)	Zeller et al.	3
ARHGEF17	ENST00000263674					
FAM168A	ENST00000064778	ILMN_1847308	blood	1.28×10^{-6} (p-value)	Westra et al.	7

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000114360	1	ENCP00000013663	RP11-800A3.2
		ENCP00000013698	RAB6A
		ENCP00000013679	RELT
		ENCP00000013669	P2RY6
		ENCP00000013664	P2RY2
		ENCP00000013678	RELT
		ENCP00000013683	RP11-809N8.4
ENCE00000114413	1	ENCP00000013676	ARHGEF17
		ENCP00000013668	P2RY6
		ENCP00000013686	RP11-809N8.5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001605004 (enhancer)	1	cervix (HeLa-S3)	PolII, H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K36me3, H3K4me3
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		blood (K562)	H3K79me2
		blood (DND-41)	H3K4me2, H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000565215 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H4K20me1, H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
ENSR00001605010 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		HSMMtube	H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
ENSR00000565218 (promoter flanking region)	1	NHLF	DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1
		A549	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00000565222 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
ENSR00000565232 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K4me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H2AZ, H3K4me1, H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM168A	downstream gene variant	1655	ENST00000064778	NM_001286050.1	ENSP00000064778	2
FAM168A	downstream gene variant	2305	ENST00000356467	NM_015159.2	ENSP00000348852	1
FAM168A	downstream gene variant, upstream gene variant	181	ENST00000432223	?	?	2
FAM168A	downstream gene variant	2305	ENST00000450446	NM_001286051.1	ENSP00000390501	1
HMGN2P38	downstream gene variant	1520	ENST00000423806	?	?	1
RELT	downstream gene variant	1360	ENST00000064780	NM_152222.1	ENSP00000064780	1
RELT	downstream gene variant	591	ENST00000393580	NM_032871.3	ENSP00000377207	2
RELT	upstream gene variant, downstream gene variant	587	ENST00000545886	?	?	3
RELT	downstream gene variant	2110	ENST00000537771	?	?	2
RELT	downstream gene variant	2737	ENST00000544075	?	ENSP00000440562	2
RELT	downstream gene variant	1358	ENST00000539134	?	?	1
RP11-809N8.2	upstream gene variant	795	ENST00000544674	?	?	2
	upstream gene variant, downstream gene variant	1619		?	?	3

RP11-809N8.4			ENST00000536855			
RP11-809N8.4	upstream gene variant, downstream gene variant	1619	ENST00000542598	?	?	3
RP11-809N8.5	upstream gene variant	2522	ENST00000540641	?	?	1
RP11-809N8.6	upstream gene variant, downstream gene variant	1940	ENST00000539481	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FAM168A	ENST00000064778	NM_001286050.1	ENSP00000064778	H	caT/caC	1
FAM168A	ENST00000356467	NM_015159.2	ENSP00000348852	H	caT/caC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FAM168A	ENST00000064778	NM_001286050.1	ENSP00000064778	26
FAM168A	ENST00000432223	?	?	5
FAM168A	ENST00000450446	NM_001286051.1	ENSP00000390501	26
FAM168A	ENST00000356467	NM_015159.2	ENSP00000348852	26
RELT	ENST00000393580	NM_032871.3	ENSP00000377207	3
RELT	ENST00000545687	?	ENSP00000439352	3
RELT	ENST00000544075	?	ENSP00000440562	3
RELT	ENST00000064780	NM_152222.1	ENSP00000064780	3
RP11-809N8.4	ENST00000536855	?	?	1
RP11-809N8.4	ENST00000542598	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FAM168A	ENST00000064778	NM_001286050.1	ENSP00000064778	1
RELT	ENST00000064780	NM_152222.1	ENSP00000064780	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RELT	ENST00000539134	?	1