SNiPAcard

Block annotations

Block info

genomic range	chr19:38,792,172-38,856,372
block size	64,201 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.210 [-5.585 – 0.64]	**gene(s) hit or close-by**	AC005625.1, C19orf33, CATSPERG, CTB-102L5.4, KCNK6, YIF1B
phastCons	μ = 0.005 [0 – 0.054]	**eQTL gene(s)**	C19orf33, CATSPERG, PPP1R14A, SPINT2, YIF1B
GERP++	μ = -1.340 [-8.19 – 2.29]	**potentially regulated gene(s)**	PSMD8
CADD score	μ = 3.520 [0.004 – 13.82]	**disease gene(s)**	SPINT2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SPINT2	DIARRHEA 3, SECRETORY SODIUM, CONGENITAL, WITH OR WITHOUT OTHER [...]	OMIM	MIM:270420
SPINT2	Congenital sodium diarrhea	OrphaNet	OrphaNet:103908

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C19orf33	?	ENSG00000167644	muscularis mucosae	2.60×10^{-7} (p-value)	GTEx Portal V6	19
PPP1R14A	?	ENSG00000167641	muscularis mucosae	6.19×10^{-7} (p-value)	GTEx Portal V6	20
YIF1B	?	ENSG00000167645	tibial nerve	2.84×10^{-6} (p-value)	GTEx Portal V6	17
C19orf33	?	ENSG00000167644	tibial nerve	1.90×10^{-17} (p-value)	GTEx Portal V6	28
PPP1R14A	?	ENSG00000167641	tibial nerve	2.57×10^{-5} (p-value)	GTEx Portal V6	2
YIF1B	?	ENSG00000167645	sun exposed skin	2.70×10^{-9} (p-value)	GTEx Portal V6	28
CATSPERG	?	ENSG00000099338	sun exposed skin	2.74×10^{-6} (p-value)	GTEx Portal V6	3
C19orf33	?	ENSG00000167644	left ventricle	5.01×10^{-10} (p-value)	GTEx Portal V6	28
C19orf33	?	ENSG00000167644	blood	5.45×10^{-10} (p-value)	GTEx Portal V6	28
PPP1R14A	?	ENSG00000167641	blood	9.63×10^{-6} (p-value)	GTEx Portal V6	2
CATSPERG	?	ENSG00000099338	thyroid	2.25×10^{-8} (p-value)	GTEx Portal V6	4
C19orf33	?	ENSG00000167644	subcutaneous adipocytes	6.37×10^{-11} (p-value)	GTEx Portal V6	28
C19orf33	?	ENSG00000167644	spleen	4.01×10^{-6} (p-value)	GTEx Portal V6	1
C19orf33	?	ENSG00000167644	tibial artery	5.06×10^{-6} (p-value)	GTEx Portal V6	16
SPINT2	ENST00000454580	ILMN_1800739	blood	4.75×10^{-5} (p-value)	Westra et al.	1
SPINT2	ENST00000301244					
SPINT2	ENST00000587090					

SPINT2	ENST00000585357					
PPP1R14A	?	ENSG00000167641	sigmoid colon	5.00×10-7 (p-value)	GTEx Portal V6	14
?	?	ILMN_1717793	monocyte	8.66×10-58 (p-value)	Zeller et al.	3
PPP1R14A	ENST00000587515	ILMN_1761968	monocyte	5.24×10-36 (p-value)	Zeller et al.	3
PPP1R14A	ENST00000301242					
PPP1R14A	ENST00000347262					
PPP1R14A	ENST00000591291					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000227970	1	ENCP00000024343	PSMD8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000346261 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, PolII, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	DNase1
		blood (K562)	Egr1, Max, H3K4me1, PolII, H3K36me3, DNase1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1, H3K36me3
		liver (HepG2)	H3K4me1, H3K4me2, DNase1, CTCF, FOSL2
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3, CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K4me1
		NHLF	DNase1, CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001647943 (promoter)	1	embryonic stem cell (H1ESC)	H3K4me3, TAF1, H3K4me2, SP1, USF1, DNase1
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K4me3, Gata2, Egr1, H3K27ac, PU1, H3K9ac, H3K79me2, PolII, H3K4me2, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K4me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		blood (GM12878)	H3K4me2, Tr4, PolII, H2AZ, DNase1, Egr1, H3K4me3
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K9ac, H3K4me2, DNase1, H3K4me3
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H3K4me3, H3K4me2
		blood (DND-41)	H3K4me2, H3K27ac, H3K4me3, H3K9ac
		breast (HMEC)	H3K4me2, DNase1, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K27ac, H3K4me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, DNase1
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me2, Max, H3K4me3
		A549	H3K27me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001647945 (CTCF binding site)	1	embryonic stem cell (H1ESC)	SIX5, Yy1, H3K4me2, SP1, USF1, H3K4me3, Rad21, Sin3Ak20, CTCF, DNase1
		HSMMtube	DNase1, CTCF, H3K4me2
		blood (K562)	Rad21, ELF1, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, Gabp, Yy1, THAP1, Max
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, DNase1, H3K4me2
		liver (HepG2)	Gabp, Yy1, Rad21, ELF1, H3K4me2, CTCF, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, H3K9ac, CTCF

	blood (GM12878)	H3K27ac, H3K4me2, CTCF, H3K4me3, Rad21, PolII, H2AZ, DNase1, Yy1, Gabp, ELF1
	nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
	skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
	NHLF	DNase1, H3K4me3, CTCF
	Osteobl	H2AZ, H3K4me3, H3K4me2, CTCF
	blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2
	breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, CTCF
	cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, Gabp, CTCF, DNase1
	monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
	endothelium (HUVEC)	Max, H3K4me3, PolII, CTCF, DNase1
	A549	DNase1, CTCF, H3K4me2, H3K4me3
ENSR00000346269 2 (promoter)	embryonic stem cell (H1ESC)	H3K4me3, H3K36me3, H3K27me3, Rad21, Sin3Ak20, CTCF, SIX5, Yy1, H3K4me2, SP1, USF1, DNase1
	HSMMtube	DNase1, CTCF, H3K4me2, H2AZ
	blood (K562)	H3K4me2, CTCF, DNase1, H3K4me3, Yy1, THAP1, Egr1, H3K27ac, Max, H3K9ac, Gabp, H2AZ, Rad21, ELF1
	skin (NHDF-AD)	DNase1, H3K4me2, H3K4me3, CTCF
	muscle (HSMM)	DNase1, H3K36me3, H3K4me2, H3K4me3, CTCF, H2AZ
	liver (HepG2)	Yy1, Rad21, ELF1, H3K4me2, CTCF, H3K4me3, Gabp, DNase1
	lung (IMR90)	DNase1, H3K4me2, H3K4me3, H3K9ac, CTCF
	blood (GM12878)	PolII, H2AZ, DNase1, Yy1, Gabp, ELF1, Rad21, H3K4me3, H3K27ac, H3K4me2, CTCF
	nervous (NH-A)	DNase1, H3K4me2, CTCF, H3K9ac, H3K4me3
	skin (NHEK)	H3K9ac, H3K27ac, H3K4me2, DNase1, CTCF, H3K4me3
	NHLF	CTCF, DNase1, H3K4me3
	Osteobl	H2AZ, H3K4me3, H3K4me2, CTCF
	blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
	breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
	cervix (HeLa-S3)	DNase1, CTCF, Gabp, H3K9ac, H3K4me2, H3K27ac, H3K4me3
	monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
	endothelium (HUVEC)	H3K36me3, Max, H3K4me3, PolII, CTCF, DNase1
	A549	DNase1, CTCF, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC005625.1	downstream gene variant, upstream gene variant	1676	ENST00000590304	?	?	5
C19orf33	upstream gene variant, downstream gene variant	18	ENST00000591852	?	?	4
C19orf33	upstream gene variant, downstream gene variant	1540	ENST00000301246	NM_033520.1	ENSP00000301246	3
C19orf33	upstream gene variant, downstream gene variant	1543	ENST00000588605	?	ENSP00000464940	3
C19orf33	upstream gene variant, downstream gene variant	1546	ENST00000589986	?	?	3
CATSPERG	upstream gene variant, downstream gene variant	721	ENST00000477793	?	?	5
CATSPERG	upstream gene variant, downstream gene variant	472	ENST00000470292	?	?	3
CATSPERG	upstream gene variant	2159	ENST00000489693	?	ENSP00000470225	1
CATSPERG	upstream gene variant	1443	ENST00000475646	?	?	1
CATSPERG	upstream gene variant	1892	ENST00000471517	?	ENSP00000468028	3
CATSPERG	upstream gene variant, downstream gene variant	627	ENST00000467739	?	?	3
CATSPERG	upstream gene variant	4772	ENST00000492088	?	?	1
CATSPERG	upstream gene variant	1361	ENST00000412458	?	ENSP00000395093	1
CATSPERG	upstream gene variant, downstream gene variant	1521	ENST00000488473	?	?	2
CATSPERG	upstream gene variant, downstream gene variant	2055	ENST00000409410	?	ENSP00000386950	4
CATSPERG	upstream gene variant	1834	ENST00000410018	?	ENSP00000387057	3
CATSPERG	upstream gene variant, downstream gene variant	262	ENST00000585424	?	?	2
CATSPERG	upstream gene variant	1834	ENST00000409235	NM_021185.4	ENSP00000386962	3

CATSPERG	upstream gene variant	1877	ENST00000312265	?	ENSP00000311314	3
CATSPERG	upstream gene variant, downstream gene variant	1908	ENST00000475343	?	ENSP00000467947	4
CATSPERG	upstream gene variant, downstream gene variant	409	ENST00000466060	?	?	5
CTB-102L5.4	downstream gene variant	1639	ENST00000587519	?	ENSP00000465845	2
CTB-102L5.4	downstream gene variant	1640	ENST00000591889	?	ENSP00000468040	2
KCNK6	upstream gene variant, downstream gene variant	486	ENST00000263372	NM_004823.1	ENSP00000263372	9
KCNK6	upstream gene variant, downstream gene variant	2371	ENST00000588137	?	?	6
YIF1B	downstream gene variant, upstream gene variant	861	ENST00000586319	?	?	4
YIF1B	downstream gene variant, upstream gene variant	218	ENST00000592694	?	ENSP00000466428	7
YIF1B	downstream gene variant, upstream gene variant	396	ENST00000585563	?	?	7
YIF1B	downstream gene variant, upstream gene variant	339	ENST00000329420	NM_001039671.2, NM_001145461.1	ENSP00000329559	7
YIF1B	downstream gene variant, upstream gene variant	148	ENST00000591755	?	ENSP00000465446	7
YIF1B	downstream gene variant, upstream gene variant	209	ENST00000337679	NM_001145463.1	ENSP00000337411	7
YIF1B	downstream gene variant, upstream gene variant	591	ENST00000587039	?	ENSP00000465277	6
YIF1B	downstream gene variant, upstream gene variant	860	ENST00000392124	NM_033557.3	ENSP00000375971	4
YIF1B	downstream gene variant, upstream gene variant	190	ENST00000589644	?	?	7
YIF1B	downstream gene variant, upstream gene variant	333	ENST00000591784	NM_001145462.1	ENSP00000465230	7
YIF1B	downstream gene variant, upstream gene variant	879	ENST00000588002	?	ENSP00000468358	4
YIF1B	downstream gene variant, upstream gene variant	169	ENST00000339413	NM_001039673.2, NM_001039672.2	ENSP00000343435	7
YIF1B	downstream gene variant, upstream gene variant	25	ENST00000589247	?	ENSP00000468463	8
YIF1B	downstream gene variant, upstream gene variant	943	ENST00000592246	?	ENSP00000467489	4
YIF1B	downstream gene variant, upstream gene variant	369	ENST00000587361	?	?	8
YIF1B	downstream gene variant	1107	ENST00000589151	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CATSPERG	ENST00000409410	?	ENSP00000386950	T	acC/acT	1
CATSPERG	ENST00000409235	NM_021185.4	ENSP00000386962	T	acC/acT	1
YIF1B	ENST00000339413	NM_001039673.2, NM_001039672.2	ENSP00000343435	V	gtG/gtA	1
YIF1B	ENST00000588002	?	ENSP00000468358	V	gtG/gtA	1
YIF1B	ENST00000592694	?	ENSP00000466428	V	gtG/gtA	1
YIF1B	ENST00000591784	NM_001145462.1	ENSP00000465230	V	gtG/gtA	1
YIF1B	ENST00000337679	NM_001145463.1	ENSP00000337411	V	gtG/gtA	1
YIF1B	ENST00000329420	NM_001039671.2, NM_001145461.1	ENSP00000329559	V	gtG/gtA	1
YIF1B	ENST00000392124	NM_033557.3	ENSP00000375971	V	gtG/gtA	1
YIF1B	ENST00000592246	?	ENSP00000467489	V	gtG/gtA	1
YIF1B	ENST00000591755	?	ENSP00000465446	V	gtG/gtA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CATSPERG	ENST00000409410	?	ENSP00000386950	10
CATSPERG	ENST00000489693	?	ENSP00000470225	6
CATSPERG	ENST00000585424	?	?	3
CATSPERG	ENST00000412458	?	ENSP00000395093	8
CATSPERG	ENST00000312265	?	ENSP00000311314	11
CATSPERG	ENST00000466060	?	?	3
CATSPERG	ENST00000409235	NM_021185.4	ENSP00000386962	11
CATSPERG	ENST00000410018	?	ENSP00000387057	11
CATSPERG	ENST00000475343	?	ENSP00000467947	3
CATSPERG	ENST00000471517	?	ENSP00000468028	11
CATSPERG	ENST00000470292	?	?	3
CTB-102L5.4	ENST00000591889	?	ENSP00000468040	2
CTB-102L5.4	ENST00000587519	?	ENSP00000465845	2
KCNK6	ENST00000588137	?	?	2
KCNK6	ENST00000263372	NM_004823.1	ENSP00000263372	2
YIF1B	ENST00000587039	?	ENSP00000465277	4
YIF1B	ENST00000587361	?	?	2
YIF1B	ENST00000586319	?	?	1
YIF1B	ENST00000339413	NM_001039673.2, NM_001039672.2	ENSP00000343435	3
YIF1B	ENST00000592694	?	ENSP00000466428	3
YIF1B	ENST00000591784	NM_001145462.1	ENSP00000465230	3
YIF1B	ENST00000337679	NM_001145463.1	ENSP00000337411	3
YIF1B	ENST00000329420	NM_001039671.2, NM_001145461.1	ENSP00000329559	3
YIF1B	ENST00000589644	?	?	2
YIF1B	ENST00000589151	?	?	1
YIF1B	ENST00000392124	NM_033557.3	ENSP00000375971	1
YIF1B	ENST00000592246	?	ENSP00000467489	1
YIF1B	ENST00000589247	?	ENSP00000468463	2
YIF1B	ENST00000591755	?	ENSP00000465446	2
YIF1B	ENST00000585563	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CATSPERG	ENST00000412458	?	ENSP00000395093	1
CATSPERG	ENST00000312265	?	ENSP00000311314	1
CATSPERG	ENST00000471517	?	ENSP00000468028	1
KCNK6	ENST00000263372	NM_004823.1	ENSP00000263372	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

C19orf33	ENST00000301246	NM_033520.1	ENSP00000301246	1
C19orf33	ENST00000588605	?	ENSP00000464940	1
CATSPERG	ENST00000409410	?	ENSP00000386950	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C19orf33	ENST00000589986	?	1
CATSPERG	ENST00000492088	?	1
CATSPERG	ENST00000585424	?	1
CATSPERG	ENST00000470292	?	1
YIF1B	ENST00000586319	?	1
YIF1B	ENST00000589644	?	1
YIF1B	ENST00000589151	?	1
YIF1B	ENST00000585563	?	1