Locus 350 (rs2227955)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,777,157-31,834,197
block size	57,041 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.384 [-1.518 – 6.28]	gene(s) hit or close-by	C6orf48, HSPA1A, HSPA1L, LSM2, NEU1, SLC44A4, SNORD48, SNORD52
phastCons	μ = 0.170 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -1.083 [-8.35 – 4.79]	potentially regulated gene(s)	AGER, AGER, AGER, AGER, AGER, AGER, AGER, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CFB, CFB, CFB, CFB, CFB, CFB, CFB
CADD score	μ = 4.804 [0.077 – 21.2]	disease gene(s)	, , , , , CFB, NEU1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
NEU1	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578
NEU1	Juvenile sialidosis type 2	OrphaNet	OrphaNet:93399
NEU1	Sialidosis type 1	OrphaNet	OrphaNet:812
NEU1	Congenital sialidosis type 2	OrphaNet	OrphaNet:93400
NEU1	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550

Direct effect on transcript

Amino acid sequence alteration

gene	effect	affected	RefSeq id	protein	exchanged	exchanged	SIFT	PolyPhen	variant(s)

	type	transcript			AA's	codons	prediction	prediction	
HSPA1L	missense variant	ENST00000375654	NM_005527.3	ENSP00000364805	A/E	gCa/gAa	?	?	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048116		C6orf48
		C6orf48
		C6orf48
		C6orf48

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445257	1	ENCP00000048184	AGER
			AGER
			AGER
			AGER
			AGER
			AGER
			AGER
		ENCP00000048138	CFB
			CFB
			CFB
			CFB
			CFB
			CFB
			CFB
			CFB

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000662343 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me2, SP1, Srf, H3K4me3, TAF1, H3K9ac, H3K27ac, H3K36me3, H3K27me3, PolII, Nrsf, Sin3Ak20
		HSMMtube	H3K27ac, H3K4me2, DNase1
		blood (K562)	Nrsf, Cfos, PolII, GTF2B, H4K20me1, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3, Ini1, Brg1, Egr1, H3K27ac, Max, ZBTB7A, H3K9ac, Gabp, Cmyc, H3K79me2, HEY1, H2AZ, TAF1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	Nrsf, PolII, H2AZ, DNase1, Egr1, Yy1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H3K4me3, H3K4ac, H3K9ac, H3K56ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, Nrsf, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000662344 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K4me3, H3K27me3, CTCF, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, Yy1, HDAC2, Tcf12, TAF1, H3K4me2, SP1, H3K36me3, H3K9ac
		HSMMtube	H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, CTCF, DNase1
		blood (K562)	H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, Brg1, Egr1, H3K27ac, Max, TAF7, H3K4me1, E2F6, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, PolII, GTF2B, Ini1, Yy1, H4K20me1
		skin (NHDF-AD)	H3K36me3, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H4K20me1, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1

liver (HepG2)	DNase1, Yy1, H4K20me1, PolII, H3K79me2, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3
lung (IMR90)	H3K4ac, H3K9ac, CTCF, H3K4me3, H3K36me3, H3K4me2, DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac
blood (GM12878)	H3K4me2, H3K9ac, H3K36me3, DNase1, TAF1, PolII, H2AZ, CTCF, Yy1, Rad21, H3K79me2, H3K4me3, H3K27ac
nervous (NH-A)	DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
skin (NHEK)	H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3, CTCF
blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
breast (HMEC)	H3K4me2, H3K36me3, H3K9ac, H3K4me3, H3K27ac
cervix (HeLa-S3)	DNase1, PolII, Cmyc, CTCF, H3K36me3, H3K79me2, H3K4me3, H3K9ac, H3K4me2, H3K27ac, TAF1, Max
monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
endothelium (HUVEC)	H3K36me3, CTCF, DNase1, PolII, H3K27ac, H3K9ac, Max, H3K4me2, H3K4me3
A549	H3K4me3, H3K4me2, H3K9ac, H3K36me3, H3K27ac, DNase1, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C6orf48	upstream gene variant	447	ENST00000395788	?	ENSP00000379134	1
HSPA1A	upstream gene variant	377	ENST00000608703	?	ENSP00000477378	2
HSPA1A	upstream gene variant	298	ENST00000375651	NM_005345.5	ENSP00000364802	2
HSPA1L	downstream gene variant, upstream gene variant	101	ENST00000375654	NM_005527.3	ENSP00000364805	2
LSM2	upstream gene variant	2528	ENST00000470086	?	?	2
LSM2	upstream gene variant	2563	ENST00000470083	?	?	2
LSM2	upstream gene variant	2396	ENST00000375661	NM_021177.4	ENSP00000364813	2
LSM2	upstream gene variant	2720	ENST00000493387	?	?	2
LSM2	upstream gene variant	2778	ENST00000475835	?	?	2
LSM2	upstream gene variant	2721	ENST00000491421	?	?	2
LSM2	upstream gene variant	2414	ENST00000477182	?	?	2
NEU1	downstream gene variant, upstream gene variant	3514	ENST00000375631	NM_000434.3	ENSP00000364782	3
NEU1	downstream gene variant, upstream gene variant	2723	ENST00000495807	?	?	3
NEU1	downstream gene variant, upstream gene variant	3615	ENST00000480384	?	?	3
NEU1	downstream gene variant, upstream gene variant	3598	ENST00000491768	?	ENSP00000433127	3
SLC44A4	upstream gene variant	1740	ENST00000487680	?	?	1
SLC44A4	downstream gene variant	3176	ENST00000475563	?	?	1
SNORD48	downstream gene variant	1070	ENST00000364953	?	?	1
SNORD52	upstream gene variant	680	ENST00000364884	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C6orf48	ENST00000375640	?	ENSP00000364791	1
C6orf48	ENST00000375638	NM_001040437.2, NM_001287486.1, NM_001287482.1, NM_001287485.1	ENSP00000364789	1
C6orf48	ENST00000614363	NM_001040438.2, NM_001287484.1	ENSP00000480389	1
C6orf48	ENST00000375635	NM_001287487.1, NM_001287488.1	ENSP00000364786	1
C6orf48	ENST00000375641	?	ENSP00000364792	1
C6orf48	ENST00000375639	NM_001287483.1	ENSP00000364790	1

C6orf48	ENST00000375633	?	ENSP00000364784	1
HSPA1L	ENST00000375654	NM_005527.3	ENSP00000364805	1
SLC44A4	ENST00000229729	NM_025257.2	ENSP00000229729	1
SLC44A4	ENST00000544672	NM_001178045.1	ENSP00000444109	1
SLC44A4	ENST00000414427	?	ENSP00000398901	1
SLC44A4	ENST00000479777	?	?	1
SLC44A4	ENST00000375562	NM_001178044.1	ENSP00000364712	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C6orf48	ENST00000395789	?	ENSP00000379135	1
C6orf48	ENST00000375642	?	ENSP00000364793	1