Locus 215 (rs10895987)

SNiPAcard

Block annotations

Block info

genomic range	chr11:64,876,740-64,936,719
block size	59,980 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.540 [-6.639 – 1.312]	gene(s) hit or close-by	AP003068.18, AP003068.9, HIGD1AP10, MIR6751, MRPL49, PDCL2P2, RNU2-23P, SPDYC, SYVN1, TM7SF2, VPS51
phastCons	μ = 0.041 [0 – 0.105]	eQTL gene(s)	ARL2, NAALADL1, SPDYC
GERP++	μ = -1.022 [-10.4 – 0.632]	potentially regulated gene(s)	CDCA5, MEN1, MEN1, SAC3D1, SIPA1, SYVN1
CADD score	μ = 4.642 [0.032 – 12.66]	disease gene(s)	MEN1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MEN1	MULTIPLE ENDOCRINE NEOPLASIA, TYPE I	OMIM	MIM:131100
MEN1	HYPERPARATHYROIDISM 1	OMIM	MIM:145000
MEN1	Zollinger-Ellison syndrome	OrphaNet	OrphaNet:913
MEN1	Familial parathyroid adenoma	OrphaNet	OrphaNet:99877
MEN1	Multiple endocrine neoplasia, type 1	OrphaNet	OrphaNet:652
MEN1	Familial isolated hyperparathyroidism	OrphaNet	OrphaNet:99879

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPDYC	?	ENSG00000204710	lung	2.92×10-6 (p-value)	GTEx Portal V6	10
SPDYC	?	ENSG00000204710	blood	2.22×10-14 (p-value)	GTEx Portal V6	11
NAALADL1	ENST00000526516	ILMN_1770963	blood	9.11×10-6 (p-value)	Westra et al.	7
NAALADL1	ENST00000532802					
NAALADL1	ENST00000528977					
NAALADL1	ENST00000533340					
NAALADL1	ENST00000531746					
NAALADL1	ENST00000356632					
NAALADL1	ENST00000534568					
NAALADL1	ENST00000530995					
NAALADL1	ENST00000533842					
NAALADL1	ENST00000532450					
NAALADL1	ENST00000533753					
NAALADL1	ENST00000358658					
NAALADL1	ENST00000355721					
NAALADL1	ENST00000340252					
NAALADL1	ENST00000529274					
ARL2	ENST00000529254	ILMN_1787879	blood	2.01×10-5 (p-value)	Westra et al.	7

ARL2	ENST00000533729					
ARL2	ENST00000529384					
ARL2	ENST00000246747					
?	?	ILMN_1777476	monocyte	2.43×10-22 (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000109797	1	ENCP00000012907	CDCA5
		ENCP00000012927	SYVN1
		ENCP00000013012	SIPA1
		ENCP00000012901	SAC3D1
		ENCP00000012872	MEN1 MEN1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000563760 (open chromatin region)	1	blood (GM12878)	DNase1
ENSR00000563763 (promoter flanking region)	1	endothelium (HUVEC)	DNase1, PolII, H3K27ac, Max, Cjun
		liver (HepG2)	H2AZ
		HSMMtube	H2AZ
		lung (IMR90)	H4K5ac
		blood (GM12878)	PolII, H2AZ
		blood (K562)	Max, H3K9ac, H2AZ, H3K4me3, PolII, H3K4me2, H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP003068.18	downstream gene variant	4869	ENST00000534819	?	?	1
AP003068.9	downstream gene variant	1446	ENST00000528887	?	?	1
HIGD1AP10	upstream gene variant	1104	ENST00000527837	?	?	1
MIR6751	upstream gene variant	776	ENST00000610479	?	?	1
MRPL49	downstream gene variant	3504	ENST00000531705	?	ENSP00000436740	1
MRPL49	downstream gene variant	3391	ENST00000532671	?	?	1
MRPL49	downstream gene variant	3391	ENST00000526319	?	ENSP00000434190	1
MRPL49	downstream gene variant	4576	ENST00000533943	?	ENSP00000434052	1
MRPL49	downstream gene variant	3383	ENST00000279242	NM_004927.3	ENSP00000279242	1
MRPL49	downstream gene variant	4452	ENST00000528529	?	?	1
MRPL49	downstream gene variant	3398	ENST00000524482	?	?	1
MRPL49	downstream gene variant	4750	ENST00000534078	?	ENSP00000434222	1
PDCL2P2	downstream gene variant, upstream gene variant	274	ENST00000533651	?	?	5
RNU2-23P	upstream gene variant	508	ENST00000410545	?	?	1
SPDYC	upstream gene variant	906	ENST00000377185	NM_001008778.1	ENSP00000366390	1
SYVN1	upstream gene variant	768	ENST00000530451	?	?	1
SYVN1	upstream gene variant	2950	ENST00000449943	?	?	1
SYVN1	downstream gene variant, upstream gene variant	1873	ENST00000525874	?	?	2
SYVN1	downstream gene variant, upstream gene variant	1941	ENST00000533685	?	?	2

SYVN1	upstream gene variant	2905	ENST00000294256	NM_032431.2	ENSP00000294256	1
SYVN1	downstream gene variant, upstream gene variant	241	ENST00000532771	?	ENSP00000432166	2
SYVN1	upstream gene variant	2904	ENST00000377190	NM_172230.2	ENSP00000366395	1
SYVN1	downstream gene variant, upstream gene variant	217	ENST00000531018	?	ENSP00000431215	2
SYVN1	upstream gene variant	2957	ENST00000307289	?	ENSP00000302035	1
SYVN1	downstream gene variant, upstream gene variant	520	ENST00000528487	?	ENSP00000431720	2
SYVN1	upstream gene variant	3008	ENST00000526060	?	ENSP00000436984	1
SYVN1	upstream gene variant	465	ENST00000527765	?	?	1
SYVN1	downstream gene variant, upstream gene variant	378	ENST00000529207	?	?	2
SYVN1	downstream gene variant, upstream gene variant	378	ENST00000527142	?	ENSP00000432888	2
TM7SF2	upstream gene variant	3003	ENST00000530892	?	?	1
TM7SF2	upstream gene variant	2610	ENST00000529601	?	ENSP00000435458	1
TM7SF2	upstream gene variant	2634	ENST00000530650	?	?	1
TM7SF2	upstream gene variant	2780	ENST00000524690	?	ENSP00000434598	1
TM7SF2	upstream gene variant	2730	ENST00000530750	?	ENSP00000432413	1
TM7SF2	upstream gene variant	2577	ENST00000526809	?	ENSP00000432171	1
TM7SF2	upstream gene variant	4241	ENST00000528802	?	ENSP00000436233	1
TM7SF2	upstream gene variant	2604	ENST00000534371	?	ENSP00000432187	1
TM7SF2	upstream gene variant	3946	ENST00000534667	?	?	1
TM7SF2	upstream gene variant	2616	ENST00000345348	NM_001277233.1	ENSP00000329520	1
TM7SF2	upstream gene variant	4282	ENST00000526048	?	?	1
TM7SF2	upstream gene variant	2622	ENST00000529414	?	ENSP00000433275	1
TM7SF2	upstream gene variant	3471	ENST00000527851	?	?	1
TM7SF2	upstream gene variant	4228	ENST00000527968	?	ENSP00000431685	1
TM7SF2	upstream gene variant	2618	ENST00000531321	?	ENSP00000431300	1
TM7SF2	upstream gene variant	2611	ENST00000525385	?	ENSP00000433325	1
TM7SF2	upstream gene variant	2621	ENST00000529292	?	ENSP00000436370	1
TM7SF2	upstream gene variant	2599	ENST00000524986	?	ENSP00000435972	1
TM7SF2	upstream gene variant	2607	ENST00000533646	?	?	1
TM7SF2	upstream gene variant	3437	ENST00000529233	?	?	1
TM7SF2	upstream gene variant	2604	ENST00000528026	?	?	1
TM7SF2	upstream gene variant	4103	ENST00000532328	?	?	1
TM7SF2	upstream gene variant	2623	ENST00000526085	?	ENSP00000434447	1
TM7SF2	upstream gene variant	2586	ENST00000279263	NM_003273.3	ENSP00000279263	1
TM7SF2	upstream gene variant	3376	ENST00000531029	?	?	1
TM7SF2	upstream gene variant	2604	ENST00000533766	?	?	1
VPS51	upstream gene variant	50	ENST00000534591	?	?	1
VPS51	downstream gene variant	918	ENST00000534124	?	ENSP00000436867	1
VPS51	downstream gene variant	869	ENST00000529180	?	ENSP00000435245	1
VPS51	downstream gene variant	774	ENST00000534557	?	ENSP00000435691	1
VPS51	downstream gene variant	949	ENST00000530773	?	ENSP00000434138	1

VPS51	downstream gene variant	1640	ENST00000528588	?	ENSP00000436134	1
VPS51	upstream gene variant	23	ENST00000533827	?	ENSP00000434542	1
VPS51	downstream gene variant	1383	ENST00000526578	?	ENSP00000431188	1
VPS51	upstream gene variant	1214	ENST00000530673	?	ENSP00000431317	1
VPS51	downstream gene variant	1541	ENST00000533656	?	?	1
VPS51	downstream gene variant	1248	ENST00000533487	?	ENSP00000433579	1
VPS51	upstream gene variant	76	ENST00000526856	?	ENSP00000435629	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
SYVN1	ENST00000526060	?	ENSP00000436984	S	tcA/tcG	1
SYVN1	ENST00000377190	NM_172230.2	ENSP00000366395	S	tcA/tcG	1
SYVN1	ENST00000294256	NM_032431.2	ENSP00000294256	S	tcA/tcG	1
SYVN1	ENST00000307289	?	ENSP00000302035	S	tcA/tcG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
VPS51	ENST00000279281	NM_013265.3	ENSP00000279281	1
VPS51	ENST00000527646	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SYVN1	ENST00000526121	?	1
SYVN1	ENST00000449943	?	1
VPS51	ENST00000531146	?	1