Locus 332 (rs2255336)

SNiPAcard

Block annotations

Block info

genomic range	chr12:10,517,808-10,644,573
block size	126,766 bp
variant count	73 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.740 [-4.39 – 0.939]	**gene(s) hit or close-by**	KLRC1, KLRC2, KLRC3, KLRC4, KLRC4-KLRK1, KLRK1, RP11-277P12.20, RP11-277P12.6, RP11-277P12.9
phastCons	μ = 0.054 [0 – 0.865]	**eQTL gene(s)**	GABARAPL1, KLRC1, KLRC2, KLRC3, KLRC4-KLRK1, KLRD1, KLRK1, RP11-277P12.10, RP11-277P12.20, TAS2R10, YBX3
GERP++	μ = -0.537 [-6.29 – 1.55]	**potentially regulated gene(s)**	CLEC1A, KLRC2, KLRC3, KLRD1, KLRK1
CADD score	μ = 3.036 [0.042 – 10.47]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Obesity-related traits	$<6.00\times10^{-6}$	GWAS Catalog	23251661	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM099345	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KLRC4-KLRK1	missense variant	ENST00000616233	NM_001199805.1	ENSP00000480609	T/A	Aca/Gca	tolerated	benign	1
KLRK1	missense variant	ENST00000396451	?	ENSP00000379726	T/A	Aca/Gca	tolerated	?	1
KLRK1	missense variant	ENST00000240618	NM_007360.3	ENSP00000240618	T/A	Aca/Gca	tolerated	benign	1
KLRK1	missense variant	ENST00000540818	?	ENSP00000446003	T/A	Aca/Gca	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KLRC2	?	ENSG00000205809	pancreas	1.05×10^{-6} (p-value)	GTEx Portal V6	36
GABARAPL1	ENST00000539170	ILMN_2151281	blood	8.21×10^{-18} (p-value)	MuTHER consortium	18
GABARAPL1	ENST00000541960					
GABARAPL1	ENST00000540424					
GABARAPL1	ENST00000266458					

GABARAPL1	ENST00000542722					
GABARAPL1	ENST00000546017					
GABARAPL1	ENST00000535576					
KLRD1	ENST00000381908	ILMN_1799134	blood	1.75×10-5 (p-value)	MuTHER consortium	14
KLRD1	ENST00000336164					
KLRD1	ENST00000344825					
KLRD1	ENST00000539374					
KLRD1	ENST00000544747					
KLRD1	ENST00000350274					
KLRD1	ENST00000543777					
KLRD1	ENST00000540271					
KLRD1	ENST00000539792					
KLRC2	?	ENSG00000205809	muscularis mucosae	2.54×10-10 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	lung	9.82×10-20 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	transformed fibroblasts	1.20×10-11 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	blood	3.58×10-13 (p-value)	GTEx Portal V6	68
KLRC3	?	ENSG00000205810	blood	2.21×10-6 (p-value)	GTEx Portal V6	15
KLRC2	?	ENSG00000205809	breast	1.41×10-9 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	thyroid	1.32×10-11 (p-value)	GTEx Portal V6	68
RP11-277P12.20	?	ENSG00000245648	thyroid	3.93×10-6 (p-value)	GTEx Portal V6	1
KLRC2	?	ENSG00000205809	transverse colon	1.12×10-8 (p-value)	GTEx Portal V6	60
KLRK1	?	ENSG00000213809	blood	5.02×10-3 (q-value)	SeeQTL DB (HapMap)	5
RP11-277P12.10	?	ENSG00000256288	sun exposed skin	4.53×10-7 (p-value)	GTEx Portal V6	40
RP11-277P12.20	?	ENSG00000245648	sun exposed skin	3.53×10-7 (p-value)	GTEx Portal V6	44
RP11-277P12.20	?	ENSG00000245648	unexposed skin	4.51×10-10 (p-value)	GTEx Portal V6	67
KLRC2	?	ENSG00000205809	subcutaneous adipocytes	2.11×10-8 (p-value)	GTEx Portal V6	62
KLRC2	?	ENSG00000205809	terminal ileum	1.84×10-8 (p-value)	GTEx Portal V6	24
KLRC2	?	ENSG00000205809	stomach	1.10×10-15 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	spleen	7.02×10-11 (p-value)	GTEx Portal V6	64
KLRC2	?	ENSG00000205809	nucleus accumbens	7.35×10-9 (p-value)	GTEx Portal V6	55
KLRC2	?	ENSG00000205809	vagina	1.21×10-9 (p-value)	GTEx Portal V6	60
KLRC2	?	ENSG00000205809	esophagus mucosa	3.88×10-10 (p-value)	GTEx Portal V6	68
RP11-277P12.20	?	ENSG00000245648	esophagus mucosa	1.10×10-27 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	anterior cingulate cortex	2.84×10-11 (p-value)	GTEx Portal V6	68
RP11-277P12.20	?	ENSG00000245648	testis	2.48×10-11 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	testis	1.63×10-6 (p-value)	GTEx Portal V6	19
KLRC2	?	ENSG00000205809	hypothalamus	4.78×10-9 (p-value)	GTEx Portal V6	56
KLRC2	?	ENSG00000205809	frontal cortex	1.07×10-13 (p-value)	GTEx Portal V6	68

KLRC4-KLRK1	ENST00000591546	1555691_a_at	blood	1.00×10-9 (p-value)	Dixon et al.	2
KLRC4-KLRK1	ENST00000586581					
KLRC4-KLRK1	ENST00000543572					
KLRC4-KLRK1	ENST00000590131					
KLRC4-KLRK1	ENST00000543812					
KLRC4-KLRK1	ENST00000585507					
KLRK1	ENST00000540818					
KLRC4-KLRK1	ENST00000539370					
KLRK1	ENST00000544449					
KLRK1	ENST00000240618					
KLRC4-KLRK1	ENST00000588263					
KLRC4-KLRK1	ENST00000539300					
KLRK1	ENST00000540267					
KLRC4-KLRK1	ENST00000616233					
KLRC4-KLRK1	ENST00000585711					
KLRC4-KLRK1	ENST00000591937					
KLRC4-KLRK1	ENST00000588447					
KLRK1	ENST00000396451					
KLRC4-KLRK1	ENST00000590323					
KLRC4-KLRK1	ENST00000591546	205821_at	blood	1.90×10-12 (p-value)	Dixon et al.	2
KLRC4-KLRK1	ENST00000586581					
KLRC4-KLRK1	ENST00000543572					
KLRC4-KLRK1	ENST00000585711					
KLRC4-KLRK1	ENST00000590131					
KLRC4-KLRK1	ENST00000543812					
KLRK1	ENST00000544449					
KLRK1	ENST00000240618					
KLRC4-KLRK1	ENST00000539300					
KLRK1	ENST00000540267					
KLRC4-KLRK1	ENST00000616233					
KLRC2	?	ENSG00000205809	tibial nerve	2.43×10-9 (p-value)	GTEx Portal V6	57
YBX3	ENST00000279550	ILMN_1782788	monocyte	1.40×10-6 (p-value)	Fairfax et al.	2
YBX3	ENST00000540447					
YBX3	ENST00000228251					
YBX3	ENST00000366286					
YBX3	ENST00000540975					
YBX3	ENST00000536823					
YBX3	ENST00000546164					
KLRC2	?	ENSG00000205809	cortex	1.77×10-7 (p-value)	GTEx Portal V6	8
KLRC2	?	ENSG00000205809	cerebellar hemisphere	3.01×10-6 (p-value)	GTEx Portal V6	4
KLRC2	?	ENSG00000205809	hippocampus	7.43×10-8 (p-value)	GTEx Portal V6	14
?	?	ILMN_1707328	liver	9.23×10-13 (p-value)	Innocenti et al.	1
KLRC2	?	ENSG00000205809	cerebellum	2.71×10-7 (p-value)	GTEx Portal V6	29
KLRC2	?	ENSG00000205809	putamen	7.07×10-7 (p-value)	GTEx Portal V6	3
KLRC1	ENST00000347831	ILMN_1752647	blood	5.64×10-5 (p-value)	Westra et al.	1
KLRC1	ENST00000359151					
KLRC1	ENST00000544822					
KLRC1	ENST00000536188					
KLRC1	ENST00000408006					

KLRC2	?	ENSG00000205809	visceral adipocytes	1.04×10-6 (p-value)	GTEx Portal V6	23
TAS2R10	?	ENSG00000121318	EBV lymphocytes	1.55×10-6 (p-value)	GTEx Portal V6	1
KLRC2	?	ENSG00000205809	caudate basal ganglia	3.48×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000133909	1	ENCP00000016103	CLEC1A
ENCE00000133997	1	ENCP00000016122	KLRK1
		ENCP00000016121	KLRK1
		ENCP00000016129	KLRC2
ENCE00000133946	1	ENCP00000016116	KLRD1
		ENCP00000016127	KLRC3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000426296 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K27ac, CTCF, Rad21, FOSL1, Yy1, TAF1, H3K4me2, SP1, H3K4me3
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	DNase1, H3K27ac, H3K9ac, H2AZ, PolII, H3K4me2, CTCF, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	CTCF, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3
		blood (GM12878)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, IRF4, DNase1, H2AZ, BCLAF1, PolII, PU1, EBF1
		lung (IMR90)	CTCF, H3K9ac, H2BK120ac, H3K4me3, H4K5ac, H3K4me2, H3K18ac, H3K27ac, H4K8ac, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, CTCF
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		cervix (HeLa-S3)	H3K9ac, DNase1, H3K4me2, H3K27ac, H3K4me3, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	PolII, H3K4me3, H3K4me2
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00001609380 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H2AZ
		blood (K562)	H3K27ac, H3K9ac, H2AZ, H3K4me3, H3K4me2, CTCF, DNase1
		skin (NHDF-AD)	H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2
		liver (HepG2)	CTCF, Rad21, H2AZ, H3K4me2, H3K4me3
		lung (IMR90)	CTCF, H4K5ac, H3K4me2, H3K18ac, H3K27ac, H4K8ac
		blood (GM12878)	H2AZ, Rad21
		nervous (NH-A)	H3K4me2
		skin (NHEK)	H3K27ac, H3K4me2, CTCF, H3K4me1
		Osteobl	H2AZ, H3K4me2, CTCF
		blood (DND-41)	H3K4me3, H3K9ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, H3K4me3, CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K4me3, H3K4me2
		A549	CTCF
ENSR00001609383 (enhancer)	1	cervix (HeLa-S3)	H3K36me3
		lung (IMR90)	H3K36me3
ENSR00001461319 (promoter flanking region)	2	cervix (HeLa-S3)	DNase1, H3K4me2, H3K36me3
		NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3

		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001609384 (CTCF binding site)	1	cervix (HeLa-S3)	DNase1, H3K4me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K27me3
		NHLF	DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000653098 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me2, H3K9ac, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K36me3, PolII, H3K9ac, DNase1, Nrsf, H3K4me2, H3K27ac, H3K4me1, TAF1, H3K4me3, H3K79me2, Gabp
		monocytes (Monocytes-CD14+)	H3K27me3, H3K27ac, DNase1
		liver (HepG2)	FOXA1
		blood (GM12878)	BCL11A, DNase1, PU1
		A549	H3K4me3, H3K4me2, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLRC1	downstream gene variant, upstream gene variant	20	ENST00000544822	NM_213658.2	ENSP00000438038	10
KLRC1	downstream gene variant, upstream gene variant	10	ENST00000347831	NM_007328.3	ENSP00000256965	10
KLRC1	downstream gene variant, upstream gene variant	560	ENST00000543893	?	ENSP00000442545	10
KLRC1	upstream gene variant	1137	ENST00000536188	NM_001304448.1	ENSP00000441432	4
KLRC1	downstream gene variant, upstream gene variant	7	ENST00000359151	NM_002259.4	ENSP00000352064	10
KLRC1	downstream gene variant, upstream gene variant	323	ENST00000537699	?	?	11
KLRC1	downstream gene variant, upstream gene variant	7	ENST00000408006	NM_213657.2	ENSP00000385304	10
KLRC2	downstream gene variant	369	ENST00000535069	?	ENSP00000438983	1
KLRC2	downstream gene variant	2105	ENST00000537017	?	ENSP00000445240	4
KLRC2	downstream gene variant	1684	ENST00000381902	NM_002260.3	ENSP00000371327	4
KLRC2	downstream gene variant, upstream gene variant	2065	ENST00000536833	?	ENSP00000444754	13
KLRC2	downstream gene variant	1694	ENST00000619500	?	ENSP00000479152	4
KLRC2	downstream gene variant	1694	ENST00000381901	?	ENSP00000371326	4
KLRC3	downstream gene variant	2288	ENST00000396439	NM_002261.2	ENSP00000379716	2
KLRC4	downstream gene variant, upstream gene variant	130	ENST00000309384	NM_013431.2	ENSP00000310216	4
KLRC4-KLRK1	downstream gene variant	3563	ENST00000616233	NM_001199805.1	ENSP00000480609	5
KLRC4-KLRK1	downstream gene variant	4201	ENST00000543812	?	ENSP00000457500	4
KLRC4-KLRK1	downstream gene variant, upstream gene variant	2121	ENST00000543572	?	ENSP00000456286	7
KLRC4-KLRK1	downstream gene variant, upstream gene variant	338	ENST00000586581	?	ENSP00000467228	5
KLRC4-KLRK1	downstream gene variant	4269	ENST00000591546	?	ENSP00000468432	3
KLRC4-KLRK1	downstream gene variant, upstream gene variant	338	ENST00000539300	?	ENSP00000455951	6
KLRC4-KLRK1	downstream gene variant	188	ENST00000585507	?	ENSP00000465434	2

KLRC4-KLRK1	downstream gene variant, upstream gene variant	69	ENST00000539370	?	?	3
KLRC4-KLRK1	downstream gene variant, upstream gene variant	2128	ENST00000585711	?	ENSP00000467316	5
KLRC4-KLRK1	downstream gene variant	4292	ENST00000590131	?	ENSP00000467152	3
KLRC4-KLRK1	downstream gene variant	39	ENST00000591937	?	ENSP00000466326	3
KLRK1	downstream gene variant, upstream gene variant	1452	ENST00000396451	?	ENSP00000379726	5
KLRK1	downstream gene variant, upstream gene variant	715	ENST00000544449	?	?	7
KLRK1	downstream gene variant, upstream gene variant	170	ENST00000540818	?	ENSP00000446003	9
KLRK1	downstream gene variant, upstream gene variant	722	ENST00000240618	NM_007360.3	ENSP00000240618	8
KLRK1	downstream gene variant, upstream gene variant	97	ENST00000540267	?	?	11
RP11-277P12.20	downstream gene variant	3880	ENST00000500682	?	?	1
RP11-277P12.6	downstream gene variant	2288	ENST00000539033	?	ENSP00000437563	2
RP11-277P12.9	upstream gene variant	4164	ENST00000535911	?	?	3

Putative effect on transcript

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
RP11-277P12.20	ENST00000500682	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLRC1	ENST00000544822	NM_213658.2	ENSP00000438038	6
KLRC1	ENST00000359151	NM_002259.4	ENSP00000352064	6
KLRC1	ENST00000536188	NM_001304448.1	ENSP00000441432	12
KLRC1	ENST00000543893	?	ENSP00000442545	2
KLRC1	ENST00000347831	NM_007328.3	ENSP00000256965	6
KLRC1	ENST00000408006	NM_213657.2	ENSP00000385304	6
KLRC2	ENST00000535069	?	ENSP00000438983	3
KLRC4-KLRK1	ENST00000588447	?	ENSP00000465254	25
KLRC4-KLRK1	ENST00000585507	?	ENSP00000465434	23
KLRC4-KLRK1	ENST00000543812	?	ENSP00000457500	25
KLRC4-KLRK1	ENST00000591546	?	ENSP00000468432	25
KLRC4-KLRK1	ENST00000590323	?	ENSP00000467880	25
KLRC4-KLRK1	ENST00000539300	?	ENSP00000455951	23
KLRC4-KLRK1	ENST00000543572	?	ENSP00000456286	23
KLRC4-KLRK1	ENST00000590131	?	ENSP00000467152	25
KLRC4-KLRK1	ENST00000586581	?	ENSP00000467228	23
KLRC4-KLRK1	ENST00000616233	NM_001199805.1	ENSP00000480609	25
KLRC4-KLRK1	ENST00000539370	?	?	22
KLRC4-KLRK1	ENST00000588263	?	ENSP00000468074	25
KLRC4-KLRK1	ENST00000591937	?	ENSP00000466326	23
KLRC4-KLRK1	ENST00000585711	?	ENSP00000467316	23

KLRK1	ENST00000396451	?	ENSP00000379726	19
KLRK1	ENST00000540267	?	?	9
KLRK1	ENST00000240618	NM_007360.3	ENSP00000240618	18
KLRK1	ENST00000544449	?	?	18
KLRK1	ENST00000540818	?	ENSP00000446003	16
RP11-277P12.20	ENST00000500682	?	?	32
RP11-277P12.6	ENST00000539033	?	ENSP00000437563	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLRC1	ENST00000544822	NM_213658.2	ENSP00000438038	2
KLRC1	ENST00000359151	NM_002259.4	ENSP00000352064	2
KLRC1	ENST00000347831	NM_007328.3	ENSP00000256965	2
KLRC1	ENST00000408006	NM_213657.2	ENSP00000385304	2
KLRC4-KLRK1	ENST00000588447	?	ENSP00000465254	1
KLRC4-KLRK1	ENST00000585507	?	ENSP00000465434	1
KLRC4-KLRK1	ENST00000543812	?	ENSP00000457500	1
KLRC4-KLRK1	ENST00000591546	?	ENSP00000468432	1
KLRC4-KLRK1	ENST00000590323	?	ENSP00000467880	1
KLRC4-KLRK1	ENST00000539300	?	ENSP00000455951	1
KLRC4-KLRK1	ENST00000543572	?	ENSP00000456286	1
KLRC4-KLRK1	ENST00000590131	?	ENSP00000467152	1
KLRC4-KLRK1	ENST00000586581	?	ENSP00000467228	1
KLRC4-KLRK1	ENST00000588263	?	ENSP00000468074	1
KLRC4-KLRK1	ENST00000585711	?	ENSP00000467316	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KLRC4-KLRK1	ENST00000539370	?	2
KLRK1	ENST00000540267	?	3
KLRK1	ENST00000544449	?	3
RP11-277P12.20	ENST00000500682	?	3