SNiPAcard

Block annotations

Block info

genomic range	chr7:23,206,329-23,321,521
block size	115,193 bp
variant count	33 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.086 [-3.04 – 1.42]	**gene(s) hit or close-by**	GPNMB, KLHL7
phastCons	μ = 0.123 [0 – 0.996]	**eQTL gene(s)**	AC005082.12
GERP++	μ = -0.219 [-6.54 – 3.09]	**potentially regulated gene(s)**	GPNMB, TOMM7
CADD score	μ = 5.205 [0.224 – 17.15]	**disease gene(s)**	KLHL7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KLHL7	RETINITIS PIGMENTOSA 42	OMIM	MIM:612943
KLHL7	Retinitis pigmentosa	OrphaNet	OrphaNet:791

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC005082.12	?	ENSG00000226816	blood	3.64×10^{-11} (p-value)	GTEx Portal V6	29
AC005082.12	?	ENSG00000226816	subcutaneous adipocytes	5.45×10^{-8} (p-value)	GTEx Portal V6	8

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000781966	1	ENST00000381990, ENST00000258733, ENST00000465673	GPNMB
FFCP00000781967	1	ENST00000381990, ENST00000258733, ENST00000465673	GPNMB

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000476393	1	ENCP00000051286	TOMM7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000065469 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2, USF1
		HSMMtube	DNase1, H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K4me2, H3K27me3
		lung (IMR90)	H3K27me3

		blood (GM12878)	H3K4me2, H3K27me3, PU1, H2AZ
		nervous (NH-A)	DNase1, H3K4me2, H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K4me2, H3K4me3, H3K36me3
		NHLF	H3K4me3, H3K27me3, DNase1
		Osteobl	H3K4me2, H2AZ, H3K27ac, H3K4me3
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K4me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K27me3
		A549	H3K27me3
ENSR00001558007 (promoter flanking region)	1	HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K36me3
ENSR00000065473 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GPNMB	downstream gene variant	1404	ENST00000409458	?	ENSP00000386476	3
GPNMB	downstream gene variant	786	ENST00000459927	?	?	3
GPNMB	downstream gene variant	889	ENST00000474157	?	?	2
GPNMB	downstream gene variant	926	ENST00000492858	?	?	3
GPNMB	upstream gene variant, downstream gene variant	1782	ENST00000492512	?	?	4
GPNMB	downstream gene variant	43	ENST00000487890	?	?	4
GPNMB	downstream gene variant	778	ENST00000465673	?	?	3
GPNMB	upstream gene variant, downstream gene variant	1455	ENST00000478451	?	?	5
GPNMB	downstream gene variant	1077	ENST00000381990	NM_001005340.1	ENSP00000371420	2
GPNMB	upstream gene variant, downstream gene variant	25	ENST00000468723	?	?	6
GPNMB	upstream gene variant, downstream gene variant	223	ENST00000479625	?	?	6
GPNMB	downstream gene variant	1076	ENST00000258733	NM_002510.2	ENSP00000258733	2
GPNMB	upstream gene variant, downstream gene variant	650	ENST00000470994	?	?	6
GPNMB	upstream gene variant, downstream gene variant	323	ENST00000463011	?	?	6

GPNMB	upstream gene variant, downstream gene variant	323	ENST00000463011	?	?	6
KLHL7	downstream gene variant	625	ENST00000469576	?	?	10
KLHL7	upstream gene variant, downstream gene variant	885	ENST00000469845	?	?	11

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
GPNMB	ENST00000409458	?	ENSP00000386476	D	gaT/gaC	1
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	D	gaT/gaC	1
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	D	gaT/gaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPNMB	ENST00000409458	?	ENSP00000386476	4
GPNMB	ENST00000487890	?	?	3
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	14
GPNMB	ENST00000479625	?	?	3
GPNMB	ENST00000492858	?	?	3
GPNMB	ENST00000492512	?	?	1
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	14
GPNMB	ENST00000459927	?	?	2
GPNMB	ENST00000470994	?	?	3
GPNMB	ENST00000465673	?	?	5
KLHL7	ENST00000521082	?	ENSP00000430351	12
KLHL7	ENST00000469576	?	?	2
KLHL7	ENST00000339077	NM_001031710.2	ENSP00000343273	12
KLHL7	ENST00000409689	NM_018846.4	ENSP00000386263	12

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPNMB	ENST00000409458	?	ENSP00000386476	1
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	2
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	2
KLHL7	ENST00000521082	?	ENSP00000430351	1
KLHL7	ENST00000339077	NM_001031710.2	ENSP00000343273	1
KLHL7	ENST00000409689	NM_018846.4	ENSP00000386263	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
GPNMB	ENST00000478451	?	1
GPNMB	ENST00000492858	?	1
GPNMB	ENST00000465673	?	1