SNiPAcard

Block annotations

Block info

genomic range	chr8:21,521,899-21,553,025
block size	31,127 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.111 [-6.714 – 0.285]	**gene(s) hit or close-by**	GFRA2
phastCons	μ = 0.001 [0 – 0.005]	**eQTL gene(s)**	GFRA2
GERP++	μ = -1.147 [-5.4 – 4.46]	**potentially regulated gene(s)**	–
CADD score	μ = 1.791 [0.058 – 4.948]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
xanthine	5.91×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GFRA2	ENST00000524240	ILMN_1656300	skin	2.81×10^{-7} (p-value)	MuTHER consortium	12
GFRA2	ENST00000306793		adipocyte	7.39×10^{-8} (p-value)	MuTHER consortium	12

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001439445 (promoter flanking region)	1	HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K27ac, H3K4me1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
ENSR00001720518 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	Rad21
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1, H3K27ac
ENSR00001387351 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1

		Osteobl	H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	Jund, DNase1
		monocytes (Monocytes-CD14+)	DNase1
		liver (HepG2)	H3K27me3
		nervous (NH-A)	DNase1
		A549	H3K27ac, DNase1, H3K4me2, H3K4me1, H3K4me3
ENSR00001439448 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		A549	H3K27ac, DNase1, H3K4me3
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GFRA2	downstream gene variant	1429	ENST00000518077	NM_001165039.1	ENSP00000429206	4
GFRA2	downstream gene variant	512	ENST00000306793	?	ENSP00000306654	6
GFRA2	downstream gene variant	848	ENST00000524240	NM_001495.4	ENSP00000428518	5
GFRA2	downstream gene variant	1337	ENST00000517328	?	ENSP00000429445	4
GFRA2	downstream gene variant	1676	ENST00000517892	NM_001165038.1	ENSP00000429979	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GFRA2	ENST00000517892	NM_001165038.1	ENSP00000429979	1
GFRA2	ENST00000517328	?	ENSP00000429445	1
GFRA2	ENST00000524240	NM_001495.4	ENSP00000428518	1
GFRA2	ENST00000518077	NM_001165039.1	ENSP00000429206	1
GFRA2	ENST00000306793	?	ENSP00000306654	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GFRA2	ENST00000517892	NM_001165038.1	ENSP00000429979	1
GFRA2	ENST00000517328	?	ENSP00000429445	1
GFRA2	ENST00000524240	NM_001495.4	ENSP00000428518	3
GFRA2	ENST00000518077	NM_001165039.1	ENSP00000429206	1
GFRA2	ENST00000306793	?	ENSP00000306654	1